Introduction to genetics and cytogenetics.

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INTRODUCTION TO

GENETICS AND CYTOGENETICS

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INTRODUCTION

T O

Genetics and

Cytogenetics

BY HERBERT PARKES RILEY

Head, Department of Botany

University of Ker^tucky

^^M^AC^

JOHN WILEY & SONS, INC., NEW YORK

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PREFACE

In this book I have endeavored to state and to explain the basic principles of biological inheritance and to show the importance of those principles to man, to the improvement of plants and animals, and to organic evolution. I have attempted to present this material in a simple fashion so that any reader can grasp the fundamentals of heredity in spite of limited biological training. However, I have also included some of the data that support these principles so that the student who wishes can acquire an adequate background for further studies in heredity, and I have added a fairly extensive bibliography so that the more serious student will have a diversified list of some of the items of the research literature should he wish more information on a subject than a book of this size can offer.

Throughout this book I have stressed general principles rather than practical applications and have drawn my illustrations

from both the Plant and the Animal Kingdoms. For the reader who is interested in human biology, references to inherited traits are numerous, and Chapters 3 and 19 should be of especial importance. The emphasis on principles and the variety of the illustrations should make this book of value to students of agriculture, psychology, and sociology. It should serve also as a foundation for advanced work in genetics and cytogenetics.

The book is readily divisible into four parts. The first five chapters provide a survey of general biological information which must be understood before progressing into the field of genetics itself. In Chapters 6 through 13 I discuss the fundamental principles of the transmission of genes. In discussing the method by which genes are distributed from generation to generation, I have used the cytological approach, describing chromosomes and their behavior at cell division and reproduction. Chapters 14 through 23 make up the third part of the book. They deal with the nature and physiology of genes and also include some topics of practical and of general interest.

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Chapters 24 through 30, the fourth and last part of the book,

deal with what are frequently called "chromosomal aberrations." If we accept an ideal concept of chromosomal behavior during cell division and reproduction and if we accept the 2n number as the ideal number of chromosomes in the animal soma or in the plant sporophyte and the n number as ideal in the plant gametophyte, any departure from these ideal conditions represents an aberration. The various types of aberrations are described in this section, and their bearing on problems of evolution is discussed. This material is often called "cytogenetics," although any correlation at all between genetic data and cytological observations should properly bear this designation.

Throughout I have tried to avoid being dogmatic on all or most controversial issues. Sometimes I have attempted to present all the important theories concerned in the explanation of certain data without expressing any preference, and on some points where I have favored one theory I have presented other theories for the student to consider.

Because of its scope, I have had to restrict the bibliography somewhat. Many important papers have had to be omitted entirely and where an author had published a series of papers on the same subject, I have listed only a few. Although I did not adhere rigidly to any rule, I frequently listed the first paper of the series and the most recent. I usually, also, included papers that contained extensive bibliographies or summarized information and those that were especially outstanding for the theories or conclusions that they presented. Even though a paper was

referred to in more than one chapter, I included it in the bibliography only once.

Several persons have read all or part of the manuscript, and to them I wish to express my deepest appreciation. However, I must emphasize that they are in no way responsible for any of the errors that may appear in the book. Professor George H. Shull of Princeton University has read and criticized the entire text in manuscript, and I am very grateful to him for many suggestions. I also wish to thank Professor P. W. Whiting of the University of Pennsylvania for his kindness in reading and criticizing the manuscript of parts of Chapters 16 and 29. Doctor Alexander Wiener of Brooklyn, New York, read the manuscript of most of Chapter 19 and made many suggestions

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that I greatly appreciate. I am grateful also to Doctor Edgar Anderson of the Missouri Botanical Garden for reading the page proof and for an important suggestion.

Many of the diagrams and illustrations are original, but in any book of a general nature it is necessary to borrow from the published works of others. I am indebted to Professor R. A. Fisher, also to Messrs. Oliver and Boyd, Ltd., Edinburgh, for

permission to reprint Table III from their book Statistical Methods for Research Workers. I also wish to express my appreciation to the University of Chicago Press for permission to use Figure 10, which had previously appeared in the Botanical Gazette, to the American Naturalist for permission to borrow Table 20, and to Scientific Agriculture for permission to reproduce Table 23. I wish also to express my sincerest thanks to all the numerous journals which gave me permission to use their material, to the many geneticists and cytologists who kindly lent me original drawings or cuts, and to those who gave me permission to redraw their published figures or to reproduce their data. Individual acknowledgments have been made in the legends of the figures or tables.

Herbert Parked Riley

University of Kentucky

Lexington, Kentucky

November, 1947

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CONTENTS

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[Begin Page: Page 1]

Chapter 1

GENETICS, CELLS, AND CHROMOSOMES

Genetics is one of the numerous branches of the biological sciences. It attempts to discover the laws which determine why certain individuals related by descent resemble one another or why they differ from one another. It is the science of heredity and it attempts to discover how and why certain resemblances "run in families" and why many differences are also found among members of the same family. It is one of the biological sciences for it includes both plants and animals in its investigations, and, especially in its more recent aspects, it borders upon physics and chemistry. It is, furthermore, a relatively new science, not estabhshed on a scientific basis before 1900.

The science of genetics is intimately related to another biological science, cytology. Cytology is a study of those minute living units, the cells, of which plants and animals are constructed. Among the many structures found in cells are certain bodies, the chromosomes, which have been shown to be of the greatest im-

portance to students of genetics because in them are located the hereditary units. In other words, the physical basis for the laws of heredity is to be found in the chromosomes; therefore, a knowledge of cytology or at least of chromosomal cytology is absolutely necessary for an understanding of the principles of the science of genetics.

The intimate relationship between the sciences of genetics and cytology was not realized during their early development. However, as more information became available in both fields of knowledge, a striking parallelism became evident which soon suggested that they were in reality not two separate studies but merely two pliases of one. Further experiments only served to corroborate this unity until it became evident that the close relationship between genetics and cytology was incontrovertible.

During the earher years of scientific investigations into the field of heredity, data were obtained by methods that are con-

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sidered purely genetical. AVhen the physical basis of genetic phenomena was realized numerous studies were undertaken using

the methods of both genetics and cytology and correlating data obtained by genetic procedures with observations determined by cytological techniques. This dual approach to the problems of heredity has given us the term cytogenetics, a term which emphasizes the correlation of information obtained by the two diverse techniques. Many of the methods of cytogenetics make use of chromosomal aberrations, for it is by an intense study of exceptional chromosomal behavior that we obtain our best information in regard to the normal conditions. Although cytogenetics is frequently concerned with aberrations, che term is a broad one and includes all situations in which data from cytology and genetics are studied with reference to each other.

A study of the chromosomes and of their behavior in related species and genera has sometimes aided in a better understanding of the evolutionary relationships of taxonomic groups. Many difficult problems of classification and of relationships have been clarified in whole or in part by supplementing taxonomic studies with those of chromosomal cytology. A study of phylogenetic relationships by the methods of both systematic botany or zoology and chromosomal cytology is sometimes called cytotaxonomy.

Resting Cells

As part of the biological background for a study of heredity we must realize that all living organisms are composed of minute structures called cells. In the higher animals and plants the body

is made up of many cells which may differ greatly in both shape and function.

When a cell is not dividing, it is usually referred to as a resting or, more properly, a metabolic cell, and it is in this condition that most of the cells of both plants and animals are to be found.

The living part of all cells, whether in the resting stage or dividing, is a very complicated mixture of a number of different chemical substances, known as protoplasm. Under the microscope, protoplasm, while alive, appears as a colorless, optically homogeneous fluid containing granules, crystals, and droplets; l)ut, when killed, fixed, and stained, it appears to have a finely

[Begin Page: Page 3]

granular nature. In the living condition protoplasm is generally considered to be an emulsion type of colloid consisting of a watery background in which are many tiny globules of an immiscible substance, giving it the appearance of milk that has been shaken up. In the watery part may be suspended many extremely small particles or granules, which may be arranged so as to form an interlacing network. In the liquid part also are various dissolved substances such as salts and sugars. Although

protoplasm is generally fluid and has a specific gravity only slightly higher than water, it may sometimes be firmer in consistency than water and more like a jelly.

Protoplasm in all typical living cells can be differentiated into two parts, the cytoplasm and the frequently more jelly-like nucleus. The outer region of the cytoplasm is firm and membranous and forms the plasma membrane. This is of great importance physiologically as it permits some substances to enter and leave the cell and prevents others from doing so. In most plant cells a cell wall surrounds the plasma membrane, but this structure is not concerned with the passage of materials into and out of the cell. In young plant cells this wall may be very thin, but in most older cells a thicker secondary wall is also present. In some types of specialized cell this secondary wall becomes very thick.

In the embryonic plant cell (Fig. 1), the space inside the cell wall is occupied by protoplasm. When the cell is not dividing, the nucleus, usually centrally located, is a round or ellipsoidal mass of protoplasm separated from the cytoplasm by the nuclear membrane, a barrier that may separate nuclear and cytoplasmic material. In the mature, unspecializecl type of plant cell known as a parenchyma cell, a large vacuole is present in the center of the cell and the cytoplasm is mostly restricted to the periphery. In the cytoplasm may be found living structures, such as the plastids and chondriosomes or mitochondria, and many non-living substances, including starch grains, protein granules, droplets of fat or oil, and various crystals.

In a typical animal cell there is no large central vacuole, and in the cytoplasm are chondriosomes and secreted granules, but no plastids. Lying in the cytoplasm to one side of the nucleus is the centrosome, a structure intimately connected with cell

[Begin Page: Page 4]

cell wall

cytoplasm

mitochondria

nuclear membrane

chromatin reticulum

nucleolus

vacuole

cell wall

chloroplast

cytoplasm

nucleus

vacuole

Fig. 1. Plant cells, (a) An embryonic cell from the root tip of an onion X2800. (6) A parenchyma cell from the mesophyll of a leaf of English ivy. X1400. Camera lucida drawings.

[Begin Page: Page 5]

division. This structure, characteristic of animal cells, is also found in some of the lower plants. The centrosome consists of a minute granule, the centriole, surrounded by a small mass of protoplasm, the centrosphere ; the protoplasm of the centrosphere is often denser than the surrounding cytoplasm. During some stages of division, star-like radiations extend outward from the centrosome into the cytoplasm, forming the aster.

Another structure characteristic of animal cells is the Golgi apparatus. It is found in the cytoplasm and frequently appears to be a system of connecting canals, but it may sometimes have a more dispersed aspect. Its function is unknown and, although it is characteristic of animal cells, it may, according to some botanists, also be present in some plant cells. No cellulose wall is present in animal cells.

Resting Nucleus

For a geneticist, the most important part of a cell is the nucleus, because in the nucleus are found the genes which determine the characteristics of the organism.

The nucleus is separated from the cytoplasm by a definite membrane, the nuclear membrane. The reality of this structure has been shown by microdissection studies. There is good evidence that this membrane is differentially permeable, as is the plasma membrane. If so, the substances to which it is impermeable may be very different from those which will not pass through the plasma membrane.

The structures inside the nuclear membrane are not easily observed in the living condition. Living nuclei generally appear clear and homogeneous, but sometimes seem to consist of many fine granules. Discerning definite structures in the nucleus is difficult because, while alive, most of the structures of a cell are colorless and have almost the same indices of refraction. Also the threads which we know to be present in the resting nucleus are extremely fine and attenuated and are, therefore, more difficult to see than during division stages, when they are many times thicker.

The structures of the nucleus are best observed if the cell is killed, fixed, and stained. By "fixing" is meant treating the

cell with certain chemicals that not only kill it but also preserve the cell structures in a condition resembling a living cell. A cell

[Begin Page: Page 6]

treated in this manner is readily stained by certain dyes, some of which stain one part of the cell and not others. The parts so stained stand out in marked contrast to the rest of the cell, and their structure is much more easily observed than it is in the living condition.

In the resting nucleus is always found the karyolymph or nuclear sap, a clear fluid consisting mainly of proteins. In fixed and stained nuclei, the nuclear membrane is generally stained but the nuclear sap appears as an unstained or very lightly stained background inside the membrane. Superimposed on this background are the chromatin reticulum and one or more nucleoli, both generally stained very deeply.

In fixed and stained slides, the chromatin reticulum has the appearance of a network and is composed of numerous very long and extremely thin threads, in loose coils. These threads are the chromonemata. When the cell divides another substance, the matrix, condenses on these threads and the chromonemata and matrix together form the chromosotnes, the most important

nuclear structures for the geneticist as they contain the genes.

In the resting nucleus, the chromosomes are individually not distinguishable, but they become identifiable as the cell divides. During cell division it is clear that they exist in definite numbers which are the same not only for all the cells of a given plant or animal, excluding certain reproductive cells, but also, with certain exceptions, for all the individuals of the same species. For example, all the somatic (that is, body or nonreproductive) cells of the fruit fly, Drosophila melanogaster, normally have 8 chromosomes, whereas those of man have 48. Similarly, cells of all normal maize plants have 20 somatic chromosomes, cells of the garden pea have 14, and those of the onion have 16.

Division of Plant Cells

All cells come from preexisting cells by division. The term cell division includes the division of both the nucleus and the cytoplasm, either of which may divide even if the division of the other does not occur. The division of the nucleus is called mitosis or karyokinesis and the division of the cytoplasm cytokinesis, but the use of mitosis for the entire process is not unknown. It is customary to divide mitosis into four or five steps which mark definite turning points in the process. Accordingly,

[Begin Page: Page 7]

these five steps are frequently recognized: propliase, prometa-phase, metaphase, anaphase, and telophase.

Prophase. During the resting or metabolic stage, the chromo-somes are so long and thin and so intertwined that they cannot be counted, and there is evidence that each is a single thread until the cell is about to begin to divide (Fig. 2). With the beginning of mitosis, however, a series of profound changes in the nature of the nucleus is begun. There are a shortening and thickening of the chromosomes and a probable loss of water and increase in staining capacity of the individual threads, and if the chromosomes in the resting nucleus are connected by branches or anastomoses, as is frequently believed, these anastomoses are withdrawn at this time. As the result of these changes the individual chromosomes are more readily seen than in the rest-ing nucleus and are no longer joined together in a reticulum. One marked feature of the chromosomes in early prophase is that they are double rather than single threads. They appear as two long threads lying parallel and close to one another, each of which contains a specialized region known as the centromere^ kinetochore, or spindle fiber attachment point. In early pro-phase, the chromosomes are still long and slender and still wind about in a number of loose coils.

As prophase progresses, the chromonemata uncoil and become thicker. The matrix begins to condense on the threads, and the

chromosomes at this stage frequently have a fuzzy outline as the result of the irregular accumulation of this deeply staining matrical material along the length of the chromosome.

The two threads that constitute the prophase chromosome are the chromatids, each consisting of a chromonema and matrix. The two chromatids are generally visibly uniform throughout except for the centromeres, and the parts on either side of the centromeres are called the arms. The region of the arm nearest the centromere is the proximal region; the part farthest away is the distal region. As prophase progresses, the matrix continues to collect around the chromatids until each chromatid is now a long, rod-like body lying next to its sister chromatid and apparently identical with it in every way. The two centromeres lie side by side and in close contact. As these changes occur in the chromosomes, the nucleolus or nucleoli get smaller and smaller and at about the end of prophase have usually com-

[Begin Page: Page 8]

[Begin Page: Page 9]

pletely disappeared. Towards the end of prophase, the chromosomes have become much shorter and thicker and stain much more deeply than in the earlier stages. They also tend to move towards the outer part of the nucleus. At this time the nuclear membrane dissolves, and with the disappearance of this boundary between the nuclear sap and the cytoplasm, prophase comes to an end.

Prometaphase. When the nuclear membrane disappears, the nuclear sap and cytoplasm are brought into direct contact, and the cytoplasm appears to act upon the nuclear sap so as to cause it to form into a long, spindle-shaped structure known as the spindle. In living cells, this structure is not easy to see, but in many fixed and stained cells it appears as a number of fine lines converging to two points. Earlier cytologists believed these lines to be fibers and regarded the spindle as composed of a number of such fibers, which were fairly widely separated in the center of the spindle but converged at the ends. This may be the correct interpretation, but the microdissection studies of Chambers have tended to show that these so-called fibers are not solid.

Whatever is the correct nature of the spindle, it is a firmer, more rigid structure than the cytoplasm in which it is embedded. If the living cells are detached from one another and mounted on a slide, the spindle is crushed only with difficulty, and the

cells generally lie so that the spindle is parallel rather than perpendicular to the surface of the slide. The spindle is of great importance in cell division and, if it fails to function properly, mitosis will be abnormal.

The spindle tapers at each end and may or may not come to a sharp point. The ends are called the poles, and the region equidistant between them, the equator. When the spindle has formed, the chromosomes released by the breakdown of the nuclear membrane move towards the equator.

Metaphase. At metaphase, the chromosomes are seen to lie on the equator of the spindle. They frequently arrange themselves so that they lie on the outer part of the spindle with only the centromeres on the equator but sometimes, especially when they are small and numerous, the chromosomes are found in the center as well as in the outer region of the spindle (Fig. 3). The centromeres always lie on the equator, forming an equatorial

[Begin Page: Page 10]

plate, and the arms often extend away from the equator and may frequently project into the cytoplasm.

The metaphase chromosomes are thick, deeply staining struc-

tures. They frequently appear as rod-shaped, V-shaped, or J-shaped bodies, and their particular appearance depends upon the location of the centromere. If it is at the end {terminal attachment) the chromosome will appear rod-shaped; if it is at or very near the center {median or submedian attachment) it is V-shaped; and if it is near but not at the end {subterminal attachment) it has the shape of the letter J. The centromere appears in the metaphase chromosome as a constriction. In addition to the centromere, secondary constrictions may be present near the end and may be very long and deep, so that the end of the chromosome appears as a little knob, called a satellite or trabant. The function of these secondary constrictions is not well known, but on some chromosomes they are regions at which the chromosome is attached to the nucleolus during the resting stage and from which the nucleolus begins to form at telophase. Each metaphase chromosome still consists of two chromatids but they are very close to one another

FiG. 3. Polar views of metaphase. (a) In the egg of the animal, Ascaris megalocephala ; X775. (6) In cells of the root tip of Iris fulva; X1500. Camera lucida drawings.

[Begin Page: Page 11]

and may be twisted about one another. They may be so close together that the separation is not visible except at the end, and the chromosome may appear as a single structure forked at the ends.

It is at metaphase that the form of the individual chromosomes is most easily seen. In some plants, as the onion, all the chromosomes are practically alike in size and shape, but in many other plants this is not so. In plants in which the chromosomes are not all alike, however, every chromosome is never different from every other one, for there are always two of each type. Thus they are always in pairs, and the two members of any pair are called homologues or homologous chromosomes. The chromosomes occur in pairs because one member of each pair has been received from the male parent and the other member from the female parent, and these two chromosomes are identical as far as visual means can detect. In the onion, for example, where all chromosomes look alike, pairs still are present but are not readily detectable because of the general morphological similarity of all the chromosomes. The onion has 16 chromosomes, but, since the chromosomes occur in pairs, it is equally accurate to say that the onion has 8 pairs. This method of designation is frequently used. Similarly, maize has 20 chromosomes or 10 pairs, rye has 14 chromosomes or 7 pairs, and cabbage has 18 chromosomes or 9 pairs.

In general, we may say that a plant has n pairs of chromosomes, or 2n chromosomes, where n is a specific number, such

as 10 for maize and 7 for some wheat species. In mitosis in most organisms, all the chromosomes are spread out at random on the equatorial plate. Even though the chromosomes exist in pairs, any one chromosome can ordinarily lie next to any other, and there is absolutely no tendency for the two members of a pair to lie near one another. This is a general rule, although there are some outstanding exceptions.

Anaphase. After the chromosomes have become arranged on the equator, the two chromatids of each chromosome move apart from one another, each going towards its nearest pole. How this movement is brought about is still a puzzle, but possibly it is initiated by a repulsion of perhaps an electrical nature between the two centromeres. The centromeres are the active force in the separation of the daughter chromosomes, and the arms are

[Begin Page: Page 12]

dragged along passively by the centromeres. After the chromatids have begun to pull apart, they are referred to as daughter chromosomes.

Anaphase begins as soon as the centromeres begin to move to the poles and ends when all the centromeres have arrived at the poles. Although the initial movement is probably always due

to the repulsion of the centromeres, the final movement is sometimes accomplished by an elongation of the central part of the spindle after the two groups of daughter chromosomes have progressed part of the way towards the poles.

Telophase. As soon as the two groups of chromosomes, with the centromeres in advance, have arrived close to the poles, a nuclear membrane begins to form around each group and finally completely encloses it. At this stage, the cell has two new nuclei, but the remains of the spindle still persist between them. At the equator, each spindle "fiber" begins to liquefy, and finally a thin fluid area is found extending completely across the cell. This plate cuts the cell into two complete halves, and where the cytoplasm comes in contact with this liquid plate, a plasma membrane is formed. The cytoplasm is now divided into two parts, and each part has a new nucleus.

The changes in the nucleus during telophase are practically the reverse of those during prophase. After the nuclear membranes have formed around the groups of daughter chromosomes, the chromosomes themselves become extremely long and thin, and are consequently less deeply stained. Part of this process is due to a loss of the matrix which had collected during prophase around the threads. The nucleolus or nucleoh reappear and become large as telophase progresses. After these changes are concluded, each new daughter nucleus resembles the resting nucleus of the original cell.

Significance of Mitosis

If the plant we were studying was the onion, there were 16 chromosomes in the resting stage' of the cell before division. At prophase, each chromosome consisted of 2 chromatids so that there were 32 chromatids. As anaphase separation took place, the 2 chromatids of each chromosome became new chromosomes so that, during anaphase, there were 32 chromosomes, 16 of which went to each daughter cell. Therefore, each new cell has

[Begin Page: Page 13]

16 chromosomes, or the same number as the parental cell. As the result of this mechanism, each cell of the body has the same number of chromosomes. These body cells, as distinguished from the reproductive cells, are called somatic cells, and ordinary mitosis of body cells is frequently called somatic mitosis to distinguish it from the type of mitosis which forms reproductive cells and which will be described in a later chapter.

Mitosis in Animals

Mitosis in the onion root tip is frequently studied in both botanical and zoological courses. It is typical of higher plants

and is fundamentally the same as in the higher animals, although

d e

Fig. 4, Mitosis in an animal cell: (a) prophase; (6) metaphase; (c) early anaphase; (d) late anaphase; (e) late telophase. Diagrammatic.

there are some differences in animal cells that should be considered. The chief difference between the two groups of organisms lies in the formation of the spindle, which arises in animal cells from two centrosomes (Fig. 4).

Chromonemata

Only an indication so far has been given of the internal structure of the chromosome. When stained with hematoxylin.

[Begin Page: Page 14]

no structure is generally visible inside the metaphase or anaphase chromosome. If, however, the cells are pretreated before fixation with weak ammonia, hot water, or several other agents, and stained with crystal violet, each chromatid is seen to consist

of a thin, coiled thread, the chromonema (Fig. 5), or gene string, surrounded by a wide matrix.

Although some visual evidence seems to indicate that the chromonemata become double during late prophase or early meta-

FiG. 5. Coiled chromonemata in Trillium, (a) Anaphase, (b) Late diakinesis showing five pairs of chromosomes, each with its four chromatids, (c) Diakinesis, showing two unpaired chromosomes; two pairs of chromosomes are at the top and one pair is at the right side; at the bottom are the paired E chromosomes, each of whose arms is as long as most of the other pairs. (Photomicrographs courtesy of Dr. C. L. Huskins.)

phase one cell division before that at which the halves separate to opposite poles, the chromosomes, when subjected to bombardment by X-rays, usually behave as a single structure at metaphase, anaphase, and telophase and in the resting stage and do not behave as a double structure until early prophase or just before prophase. Apparently the reason for this is that at metaphase and anaphase, the chromosome as a w^hole is single even though the chromonema inside may be double, and it is the whole chromosome that reacts to the X-rays.

The process by which a new chromonema forms from an old one is not adequately known, but there is some evidence that each constituent part of a chromonema, the gene, regenerates another identical with and alongside itself. These new genes then become joined up, and a duplicate of the original chro-

monema is formed.

[Begin Page: Page 16]

Chapter 2

CHROMOSOMES AND GENES

As usually observed, chromosomes appear as thick, homogeneous bodies during metaphase and anaphase, and as long, thin threads at prophase. The prophase threads do not visually show any differentiations except for the centromeres and numerous granules, or chromomeres, which are most noticeable during the prophase of the first of the two divisions that form spores in most plants and germ cells in animals. Although a chromosome shows little differentiation visually, it consists of a large number of submicroscopic structures, or genes, spaced along the thread in a linear order but not an equal distance apart from one another. The chromonema is a series of such genes separated by inert regions and has therefore been referred to as a gene string. These genes, or factors as they are sometimes called, are too small to be observed with the photomicroscope, but they are regions of great physiological activity. Exerting their effect during development and in conjunction with one another and with the environment, the genes determine the various physical and mental characteristics which make up the mature plant or animal. It is very difficult to obtain a reasonably accurate estimate of the size of a gene, but it has been suggested that it is

about the size of some viruses and that its maximum dimensions are roughly about 100 X 20 X 20 mix.*

The number of genes in a plant or animal is apparently large. Not many organisms have been studied intensively but in Drosophila melanog aster, over 500 genes have been discovered, and in Zea mays, about 400. These are by no means the total numbers of genes in these organisms but only the numbers found and identified; they are probably only small fractions of the total numbers. Muller has estimated that at least 1150 genes are

* One fi, or micron, is one one-thousandth of a millimeter. A m/i is a millimicron and is one one-thousandth of a micron.

[Begin Page: Page 17]

present in Drosophila melanog aster, and Waddington places the figure for this fruit fly and also for lilies at possibly about 10,000.

Alleles

It was pointed out in Chapter 1 that the chromosomes are

always found in pairs in typical organisms. Since each chromosome always has an identical mate or homologue, the genes must

Fig. 6. Male (left) and female of wild type of Drosophila melanogaster. Note the sex combs on the legs of the male. Xl9- Camera lucida drawings.

also always be found in pairs. For example, in the long, V-shaped chromosome of Drosophila melanogaster, known as chromosome II, a gene about one-third of the distance from one end affects the shape and size of the fly's wing. Since every normal fly of that species has two of those chromosomes, every normal fly must have two of those genes that affect the wing. In other words, on each homologous chromosome there is a gene at a particular place or locus that always affects the wing. However, the genes at that locus on the two homologues do not always affect the wing in the same manner in all flies of that species. In most flies, the two genes will be alike, and each will act to produce a normal, or wild-type, wing in the adult (Fig. 6). Adult flies possessing those two genes will have normal wings. There are other flies, however, in which the two genes

[Begin Page: Page 18]

at that same place in chromosome II will act differently on the developing wing of the fly and will produce in the adult not normal or wild-type wings, but very small, vestigial wings (Fig. 7). Flies with these tiny, undeveloped wings cannot fly but can only crawl about like ants. Obviously, such vestigial-winged flies would be at a great disadvantage in nature and would probably not survive in competition with their wild-type relatives. They have been found in laboratory stocks and have been pre-

FiG. 7. Mutant wing types of Drosophila melanogaster. Left, ^'estigial wings. Right, miniature wings. Both are female. Xl8. Camera lucida drawings.

served for many generations in an environment free from competition and in which they do not have to travel great distances in search of food.

The important thing to note is that the genes at that particular locus of the second chromosome always affect the wing, even though the effect produced is not always the same. The gene that produces a normal wing and the gene for vestigal wing cannot, therefore, be so very different. They must be much more alike than either one would be like the gene that produces white eyes or the gene that produces yellow body color or the gene that produces forked bristles, hairless body, or purple-colored eyes. They are very similar, although not identical, not so much because they affect the same part of the body as because they are at the same locus. In a sense, then, they are merely variants of

the same gene, and not two distinctly different genes. The

[Begin Page: Page 19]

"vestigial gene" can be considered just a different form of the "normal-wing gene" which is present at that locus and vice versa. Two genes at the same locus but producing somewhat different effects on the developing individual are called allelomorphs or, more usually, alleles. Therefore, the vestigial gene is an allele of the gene for normal wings which is at the same locus.

Flies with two normal-wing genes have normal wings and those with two genes for vestigial wings will have vestigial wings. Every normal fly must have two members of chromosome II, but some may have one chromosome with a gene for normal wings and the other with a gene for vestigial wings. The question may well be asked whether the wings of such flies will be normal, vestigial, intermediate, or something else. In this particular case, such an adult fly would have normal wings because it happens that the effect produced by the wild-type gene during the development of the fly completely overcomes the effect produced by the vestigial gene. Whenever one allele alone is expressed to the exclusion of the other, the allele whose effect is expressed is said to be dominant over the one whose effect is not expressed,

known as a recessive gene.

An organism in which the two genes at one locus are identical is homozygous for that gene. Thus the vestigial-winged fly and the fly with the two genes for normal wing are homozygous. The normal-winged fly that has one dominant gene and also the recessive allele for vestigial wings is heterozygous. An individual in which the two genes at any one locus are different is heterozygous for that pair of alleles.

The two members of all pairs of alleles do not show this dominant-recessive relationship for some heterozygous individuals are intermediate in nature. Although it is usually true that the wild-type or '"normal" gene is dominant over its allele, sometimes the "normal" gene is recessive to the "abnormal." Thus in Drosophila the gene for bar eyes that produces an eye in which most of the facets are undeveloped is partially dominant over the normal, and the gene for hairless body is dominant over its allele that produces the wild-type or normal hairy condition. Such traits as normal wings, vestigial wings, bar eyes, and hairless bodies are frequently referred to as characters.

[Begin Page: Page 20]

Heterozygous plants and animals show that all individuals

that look alike are not necessarily genetically alike. Both the homozygous wild-type fly and the heterozygote have perfectly normal wings and are absolutely indistinguishable in appearance. It can be shown that they are different genetically, however, when they are used to produce subsequent generations. The vestigial-winged fly, on the other hand, must have two genes for vestigial wings because if it had even one of the dominant genes for normal wings it would have normal wings. It is possible, therefore, to identify flies that are homozygous recessives by examining their external appearance, or phenotype, and thus to know their genetic constitution, or genotype. All individuals that are homozygous for a recessive gene are alike both phenotypically and genotypically (with respect to that locus). All individuals that have a dominant gene are alike phenotypically but may be different genotypically for they may be either homozygous for the dominant allele or heterozygous.

Gene Symbols

It is rather cumbersome to write "the gene for vestigial wings" and "the gene for normal wings." The geneticist, like the mathematician and the chemist, substitutes symbols for such expressions, and with a little practice such symbols are readily grasped. The gene for vestigial wings has the symbol vg, and its allele for normal wings has the symbol Vg. The small v indicates that the gene is recessive, the large V indicates a dominant gene. It happens that the allele for the vestigial gene is the one present in flies gathered in from nature; and that this gene is the one

found in such wild-type flies is indicated by research workers on Drosophila by modifying the symbol to read vg+ or +^^ or frequently by using merely a plus sign. In this book, however, the symbol Vg is generally used as it is less confusing to beginning students.

In choosing symbols to represent genes, it is helpful although not essential for the symbol to indicate the name and chief effect of the gene. The symbol vg cannot be mistaken for any gene other than vestigial, and the symbol Vg indicates clearly that this gene is the dominant allele of vestigial. However, when plants and animals were first studied genetically, symbols were assigned to genes arbitrarily. In Mendel's original paper, for

[Begin Page: Page 21]

example, the genes A and a stand, respectively, for peas with round and with wrinkled seeds; B and b symbolize genes for yellow and green cotyledons; and C and c represent genes for gray-brown and white seed coats. In organisms in which over twenty-six pairs of alleles have been discovered, the alphabet is insufficient to symbolize all the genes. It was soon recognized that various combinations of letters were necessary to provide symbols for such large numbers of genes. Thus in Drosophila melanog aster, v stands for the vermilion-eye gene, vg for the

gene for vestigial, and ve for the gene for veinlet. When this system was introduced, the second letter was written as a subscript, as Vg, Vg, Ve, and Ve, but the present system was adopted because it was easier to set in type.

Pleitropy

Since the genes Vg and vg determine the shape of the wing if all other genes that affect the wing are wild-type genes, it might be inferred that they have no effect except on the shape of the wing. This is not true for actually one gene may affect many parts of the body. Some parts are affected in a more striking manner than others, however, and a gene is usually named from the most striking effect that it produces. In Drosophila melanogaster, the gene Delta produces a delta-like expansion where the longitudinal vein of the wing joins the marginal vein; but it also produces smaller, rougher eyes, modifications of the bristles, and still other changes of an even less striking nature. It receives its name and symbol, Dl, however, from one of its most striking effects. Similarly the genes at the locus for white eye affect not only the color of the eye but also the shape and color of the spermatheca. Another rather curious case of pleiotropy, or the multiple effects of genes, is shown by the white- and red-eye genes. Flies with yellow bodies may have either red or white eyes. In the white-eyed flies, the gene for eye color affects the yellow pigment also, and affects it in such a way that it is easily extracted with alcohol.

Often cited as examples of pleiotropy are instances of pigment

formation in plants where a certain gene may produce a red pigment in several organs such as flowers, stems, and leaves. Considering these as separate organs, one may be inclined to think of the gene as simultaneously producing red flowers, red

[Begin Page: Page 22]

stems, and red leaves, and therefore as having several different effects. It is probably more correct to think of the gene as having one general effect, the production of pigment, and not several effects, for, after all, the plant is a unit and it is only the botanist who subdivides it into organs.

A very interesting situation that might readily be classed as pleiotropy if it were not sufficiently analyzed is the effect produced by the frizzle gene in poultry. This gene is an incom-

FiG. 8. Frizzle fowl. Lejt, a homozygous frizzle male. Center, homozygous frizzle female. Right, bare homozygous frizzle female. (Photographs courtesy of Dr. W. Landauer.)

pletely dominant gene. In fowl homozygous for frizzle (Fig. 8), the feathers are very abnormal, being narrow and curled and

very fragile. They break off easily, so that after a while such birds are almost featherless. In heterozygotes the feathers are wavy rather than curly, and the whole frizzled effect is less pronounced.

The frizzled and fragile condition of the feathers is apparently the only direct effect of the frizzle gene, but fowl homozygous for this gene are very different from normal fowl in many other respects. When all or most of the feathers are broken off, their insulating effect is naturally lost, and such naked fowl lose their body heat much more rapidly than normal fowl. To compensate for this excess loss of heat, a number of adaptations take place in the bodies of the frizzle individuals. Their basal metabolism is strikingly accelerated, thus increasing the supply of hormones from the thyroid and adrenal glands. This increased hormone production severely taxes these glands and results in

[Begin Page: Page 23]

changes in the glands themselves. The extent of these changes depends largely upon the temperature at which the birds are kept. If the temperature is as favorable as possible, the thyroid gland may increase in size but be otherwise normal; but if the conditions under which the fowl are kept are less favorable, the gland may be hypertrophied. If the birds are raised in a low

temperature, the loss of heat is so great and there is such a drain upon the thyroid glands that they may become exhausted and atrophied.

As a consequence of heat loss, the oxygen consumption is increased by an increase in the rate of the heart beat, accompanied by hypertrophy of the heart and an increase in the volume of the circulating blood. The excess loss of body heat also calls for a greater amount of heat production by the fowl, partly accomplished by an increase in the amount of food they eat. The increased food intake produces an enlargement of the pancreas, crop, gizzard, and kidneys. Frizzle fowl, therefore, show changes not only in their feathers but also in the nature and size of a number of their organs. All these effects are brought about because of one gene, but the effect on the feathers is the only direct effect that that particular gene has. All the other effects are indirect, not caused by the gene, although they are the result of the presence of that gene; they therefore might easily be wrongly interpreted as an example of pleiotropy. Pleiotropy refers only to the production of more than one direct effect by a gene and does not include such cases of indirect effects.

Unit Characters and Gene Interaction

We have pointed out that flies with the gene Vg have normal wings whereas those that are homozygous for vg have vestigial wings. In the earlier days of the science of genetics many similar cases were observed in which one character appeared to be due

to one gene only whereas a ^'contrasting character" was due only to the allele of that gene. The notion that a pair of contrasting characters was conditioned only by one pair of alleles led to the suggestion that an individual was made up of a large number of characters and that each one was the result of the action of one gene. Such monogenically conditioned characters were called unit characters, and an individual was thought to be a

[Begin Page: Page 24]

mosaic of these unit characters. Such a position is a very extreme one and, for most characters, is undoubtedly unsound.

In chromosome I in Drosophila melanogaster there is another pair of allelic genes which affect the shape of the wing. Flies homozygous for the gene m have miniature wings (Fig. 7) much like the wild-type wing but considerably smaller. Wild-type flies have the gene M which is dominant over the gene for miniature wings. We had previously stated that wild-type flies had the Vg gene and now we say that they have the M gene. Is this inconsistent? Actually the statements that Vg produces a wild-type wing and that M produces a wild-type wing are erroneous although such statements are frequently made by people who understand the correct situation. The wild-type wing is not produced by Vg alone or by M alone but is the result of both

genes acting together. In other words, the wild-type wing is not a unit character because it is not produced by only one gene. Similarly, the miniature wing is not the result of m alone but of m working in conjunction with Vg. Gene Vg, therefore, does not produce a normal wing; it merely produces a normal wing if M is present. Gene vg, likewise, produces a vestigial wing in the presence of M but produces a type known as vestigial-miniature if m is present. Practically, the last type is indistinguishable phenotypically from a vestigial.

The early notion of unit characters certainly cannot apply in this situation, but it is easy to see how it might have developed. If a geneticist has only wild-type and vestigial stocks of flies, he has flies which all have the M gene. Since his wild-type flies are VgVg MM genotypically and his vestigial flies are vgvg MM, the M gene is not important, and apparently wing shape is determined by Vg and vg alone. Unless he later obtained some miniature flies, the presence of gene M would never be detected and normal and vestigial wings would act simply as unit characters. If, now, another geneticist had only wild-type {VgVg MM) and miniature {VgVg mm) flies, the presence of Vg would never be revealed, and wing shape would appear to be conditioned only by the genes M and yn. If these two geneticists then got together and traded their stocks, by making the appropriate crosses they would learn of the presence of both pairs of genes and would realize that wild-type, vestigial, and miniature

[Begin Page: Page 25]

were not unit characters but were due to the interaction of two pairs of genes.

The situation is not even so simple as we have just pictured it. The wild-type wing is due not only to Vg and M but also to the alleles of the genes that produce cut wing, jammed wing, curved wing, plexus wing, curled wing, bent wing, and other wing variations. A partial formula for the wild-type wing, then, would be CtCtMM jjVgVgCC PxPxCuCuBtBt; a miniature fly would be CtCt mm ijVgVg CC PxPxCuCuBtBt] and a vestigial fly would be CtCtMM jjvgvgCC PxPxCuCuBtBt.

This example shows that the wild-type fly has a certain combination of genes. It shows further that the miniature fly differs from the wild-type in one certain pair whereas the vestigial differs from the wild-type with respect to a different pair. Ordinarily, in discussing miniature versus wild-type flies, we do not bother to write the full formula in either case but only the differential, which is M and m; we merely understand and imply that the other genes are present and are alike in each case. When we say that a vestigial fly is vgvg we recognize that these other genes are present but we omit them from the formula because they are the same in both the vestigial and wild-type flies. It is incorrect to say that Vg produces wild-type flies and vg pro-

duces vestigial, but it is permissible as a time and space saver provided we realize that a number of other genes which affect the wing are also present and that they are the same in each case.

This example of gene interaction is further interesting because it shows that the wild-type fly does not necessarily consist of all dominant genes. At the locus of jammed, the wild-type fly has the recessive gene, whereas the nonwild-type, jammed, is produced by the dominant allele. The wild-type fly has a certain combination of genes. Each other type has a somewhat different combination. Throughout the course of evolution, flies with the wild-type wing apparently were better adapted to their environment than the other types. Because of this evolutionary factor, a certain combination of genes is found much more frequently in nature than any other combination. We frequently think of this more frequent combination as the normal one because it is the one present in almost all the flies we gather in

[Begin Page: Page 26]

from nature and because the shape of the wing it produces looks more efficient and more suitable than the wing of any other gene combination.

Gene interaction is by no means confined to wing shape in Drosophila. Eye color and other traits in this fly and many characters in many other plants and animals have proved to be the result of the interaction of many genes rather than of the action of one gene alone. In fact, so many examples of interaction have been discovered that one wonders whether there are any true cases of unit characters. Certainly, in the broad sense the unit character idea is philosophically untenable for the organism is a unit and not a mosaic of independent structures. Since all the parts of a body act together for the benefit of the body as a whole, it is difficult not to believe that all the genes must act together also. Although all genes probably affect all parts of the body at least slightly or in an indirect manner, some genes affect some parts more strongly than others. In a sense, probably all genes have multiple effects and probably all characters are influenced to some degree by a number of genes.

The Genome

It has been mentioned previously that Drosophila melanogaster has eight chromosomes, or two sets of four chromosomes in its somatic cells. In fact, this is a much more significant way of stating the chromosome number. Since each chromosome has a mate, each gene must have a mate (either a similar gene or an allele). Therefore, this fly also has two sets of genes. One set is located in one set of four chromosomes, and the other set of genes is located in the other four chromosomes. Since each set of four chromosomes w^ith its set of genes is known as a genome, this particular species of fly has two genomes.

Similarly, in maize there is one set of ten chromosomes with its genes and another set of ten chromosomes which are morphologically identical with the first ten and have genes allelic to the genes of the first set. Like Drosophila melanogaster, this plant also has two genomes.

Many plants and most animals are similarly composed of two genomes, but in a number of plants and in some animals three, four, or more genomes have been found. Occasionally, also, an

[Begin Page: Page 27]

organism has been produced which has only one genome, but such organisms are usually very weak and delicate. With a few exceptions, every locus must be represented at least once for the organism to survive. If a piece of a chromosome is missing, as may result from subjecting germ cells to X-rays, a deficiency results. If the deficiency is present in the same region of two homologous chromosomes, it is homozygous, and if the deficiency is in one chromosome but not in the homologue, it is heterozygous. Heterozygous deficiencies frequently have marked phenotypic effects which are sometimes mistaken for the results of gene action. Frequently organisms with heterozygous deficiencies are less viable than those with two complete genomes, and this is

especially marked when the missing segment is a long one. AVhen the deficiency is homozygous, the organism usually fails to survive past the egg stage. In a few cases, however, where the deficiency is very short, as in the yellow deficiency of Drosophila, the organism may occasionally reach the adult stage. From these deficiencies we can conclude that, with a very few exceptions, at least one member of every pair of genes must be present for an organism to develop normally and that in most organisms two complete genomes provide the best background for normal development. Plants with more than two genomes are discussed in later sections of this book.

Multiple Alleles

In normal diploid organisms, there are two genomes and every locus is represented by two genes. It has been shown, however, that the two genes at a given locus are not necessarily alike, for in heterozygotes one is an allele of the other ; but in every heterozygote, there can be only two different alleles at any one locus.

Although any individual diploid plant or animal may have only two genes at any given locus, in many species three or more different alleles may be found at the same locus of a given chromosome distributed among the different individuals, with no individual having more than two. For example, in the common bean, some plants may be homozygous for G, a gene that determines yellow pods and green foliage, and others may be homozygous for the allele g, when they will have yellow pods and

yellow foliage. Other plants may be Gg and will look like GG plants because G is completely dominant to g. However, in some

[Begin Page: Page 28]

strains of beans, another gene, Gr, is found at the same locus. This gene produces plants with green p6ds and green foliage. At this locus, therefore, three alleles may be found, but any one bean plant can have only two of the three. More than two alleles at one locus are called multiple alleles, and series of multiple alleles are quite common among plants and animals. In beans, Gr is dominant over both G and g, and G is dominant over g. Any plant may be homozygous for any one of the three or heterozygous for any two. Thus these combinations will result:

It is customary to differentiate most of the members of a series of multiple alleles by the addition of a superscript to the symbol of the recessive although this rule has not always been adhered to. Thus at the white locus in Drosophila melanogaster, in addition to the genes w (white) and w^ or W (red or wild type), are found w'^'^, wine; w'^^, coral; w^^, blood; w^, cherry; u'°, apricot; w^, eosin; w\ ivory; w^, buff; w\ tinged; and w^^y €cru.

QUESTIONS AND PROBLEMS

1. In maize, the sugary gene su is recessive to its allele, the starchy gene Su. Would plants of these genotypes be sugary, starchy, neither, or both: Susw, susu; SuSu?

2. In four-o 'clocks, gene w produces white flowers when homozygous and gene W, its allele, produces red flowers when homozygous. There is no dominance, and the heterozygote is pink. What would be the color of plants with these genotypes : WW, ww, Ww ?

3. If you had a starchy corn plant, how could you tell whether it w^as homozygous or heterozygous? If the plant was a sugary plant could you tell?

4. Can you tell the genotypes of these plants by merely looking at the plant: white four-o'clock; sugary maize; red four-o'clock; starchy maize; pink four-o'clock?

5. Suggest workable symbols for these characters that Mendel studied:

[Begin Page: Page 29]

6. In Drosophila melanog aster , these recessive genes for eye color are found : car = carnation ; bw - brown eye ; pr - purple ; w = white ; p = pink. The dominant alleles of these genes interact to produce a wild-type (red) eye. Write the complete formula (as far as these pairs of alleles are concerned) for flies with these eye colors: pink, carnation, red, purple, white, brown.

[Begin Page: Page 30]

Chapter 3

GENES AND CHARACTERS

Genes act at various stages during the development of the organism to produce definite characters but, although each gene acts always in cooperation with other genes and with the environment, the effect of the environment may not be the same upon all genes. Some genes behave so differently in different environments that the characters they produce are strikingly different; other genes appear to produce the same result under all known environmental conditions. It cannot be too strongly emphasized that heredity and environment are factors which are continually interacting. The developed character is the product of a certain hereditary constitution and a certain set of environmental conditions both of which are acting during development to produce the character in question.

As an illustration of characters which develop in the same way under different environmental conditions and of those which are different if the environment is different, some of the genes that affect the color of the fruits in maize may be cited. Certain strains always have white ears because they have genes that produce white fruits under apparently all conditions of the environment. Other strains have genes that produce red fruits even when the plant develops under a variety of conditions, and such strains always have red-fruited ears. In some strains, however, there is a gene that produces different results, depending upon whether the ear is kept dark or is exposed to the light as it develops (Fig. 9). If the ears of plants that have this "sun-red" gene are allowed to mature normally in their husks w^here they are completely protected from sunlight, the mature ears are white. On the other hand, if the husks are removed from the developing ear, the ear turns a bright red. If only portions of the ear are exposed, only those portions will become red; the

[Begin Page: Page 31]

other parts will remain white. The sun-red gene, therefore, reacts to produce a white ear if the environment of the ear is complete darkness but produces a red ear if the ear is developed in the

Fig. 9. Red and white ears in maize. The left ear is white and the right ear is a red pericarp t3^pe. The two center ears are of the sun-red type and are white where covered and red where exposed to the sun. The red pericarp t3^pe is red whether covered or exposed. The few colored grains in the white part of the sun-red ears are the result of stray pollen. (Photographs by Dr. W. Brooks Hamilton.)

light. This is a different gene from the normal red pericarp gene, which produces red ears no matter whether the developing ear is kept light or dark.

The effect of temperature on the gene vg in Drosophila melano-

[Begin Page: Page 32]

gaster has been studied in considerable detail and is another good example of the principle that some genes produce different phenotypes if they act in different environments. Under the environment in which the flies normally develop, homozygous

vg flies have very small and very poorly developed wings when compared to those with the wild-type allele. Such flies are normally raised around "room temperature," or about 20° to 25° C. If, however, homozygous vg flies are raised during the larval stage at various temperatures from 14° to 34° C, the wings of the adult will be very different in size and shape. The wings of flies raised at 14° are smaller and are more poorly developed than those of flies raised at 22°. At increasing larval temperatures, the adult wings become increasingly larger and more and more like the wing of a wild-type, Vg, fly.

Penetrance and Expressivity

The statement made in Chapter 2, that an individual that is homozygous for a given recessive gene is phenotypically recessive, is true for most of the genes that have been studied thoroughly, but some genes prove to be exceptions. All flies of Drosophila melanogaster that are homozygous for vestigial have vestigial wings; but only about 70 per cent of the human beings that are homozygous for the recessive gene for susceptibility to poliomyelitis acquire the disease when exposed. Such genes as vg are said to have complete penetrance whereas the gene for poliomyelitis has only 70 per cent penetrance. In other words, penetrance of a gene is the percentage of all the individuals possessing that gene and showing phenotypically the character that is determined by it. If all homozygous recessives show the recessive character, the penetrance of that gene is complete. If almost but not quite all the individuals show it, the gene has a high penetrance ; but if only a small percentage of the individuals

are phenotypically recessive, the gene has a low penetrance. Dominant genes as well as recessives may differ in their penetrance.

Genes are known with various intermediate degrees of penetrance. Because genes with high penetrance are the easiest to work with, such genes have been the ones most frequently studied, but in studying human genetics many genes are encountered

[Begin Page: Page 33]

which have a low or intermediate penetrance. If we are to learn how inherited traits are transmitted in hmiian beings, these genes cannot be ignored. The analysis of poliomyelitis in McDowell County in West Virginia by Addair and Snyder points to the conclusion that this gene for susceptibility has a reduced penetrance as only 29 individuals contracted infantile paralysis whereas the relationships of the families studied would suggest that 40 individuals were homozygous for the recessive gene for susceptibility. In a case in fowl, reported by Hutt and Child, a recessive gene for inherited tremor is present in which the affected individuals continually shake to a greater or lesser extent. This gene has an unusually low penetrance. Their breeding studies indicated that about 112 individuals were homozygous recessive for this gene, but actually only 39 chicks showed the

character. Therefore, the penetrance of this gene is about 35 per cent. Genes are known with a lower penetrance than that, such as the gene for abnormal abdomen in Drosophila funehris, in which the penetrance is only 10 to 15 per cent.

It is not always clear why a given gene has a low or high penetrance. It is probably due to the nature of the gene itself. If a certain gene has low penetrance, apparently its action during development is weak and can easily be disturbed by the action of other genes and also by external factors. These genes with low penetrance are so easily affected that in most individuals their action is negatived completely and the individual develops the character of an allele. The action of genes with high or complete penetrance is so strong that in few or no cases can it be upset or blocked by any other combination of genes or by environmental conditions.

Hutt and Child interpret the low penetrance of the gene for tremor as the result of modifying genes. In some recessive individuals these modifying genes are powerful enough to prevent the recessive gene from being expressed in any degree; in other individuals some or all of these modifying genes are absent and the individuals are recessive phenotypically. In Drosophila melanogaster, the gene giant has a low penetrance, but environmental conditions are responsible. If the food is so scanty that there is extreme competition among the larvae, the action of the recessive gene for giant is inhibited in the homozygotes and the adults are normal in size.

[Begin Page: Page 34]

Thirty-nine chicks in Hutt and Child's study showed the tremor character, but the extent to which it appeared varied greatly in different individuals. In some chicks this tremor was so pronounced that they could not even stand up, whereas others showed only a barely perceptible tremor. Various intermediate conditions were observed. This difference is known as the expressivity of the character. Penetrance and expressivity are not the same thing. In determining penetrance, every chick was counted that manifested the character in any degree irrespective of the extent of the expression of the character in that individual. In determining the penetrance of the gene for poliomyelitis, Addair and Snyder also took expressivity into account. Although cases with high expressivity were readily recognized, the possibility existed that there might have been susceptible individuals in which the expressivity was so low that the infection produced only a fever or other mild illness instead of the usual crippling paralysis. A careful check was made to learn whether any brothers or sisters of paralyzed children had mild cases during the period when the less fortunate members of their family were affected. Since no such cases were found, it seemed clear that the expressivity of the gene was high and that the penetrance was not complete.

Several examples of characters produced by one or more genes have been mentioned in this and the last chapters; many more are discussed later in this book. We might mention here, however, that all organs of plants and animals are under genie control. In plants, a list of inherited characters would include stem height, length of internodes, type of branching, leaf shape, chlorophyll deficiencies, flower color and color patterns (Fig. 10), shape of flower parts, shape of fruit, color of seed coats and endosperm, and even seedlessness. In animals we could list such traits as abnormalities of bone growth in the skull and other bones (Fig. 11), the presence of excess fluid between the brain and the skull, absence or reduction of the jaws, eye color, congenital cataract, color of the fur or feathers (Fig. 12), albinism, woolly hair, hairlessness, inherited bleeding, size and weight, glandular abnormalities, and many more far too numerous to mention. Dunn and his co-workers described in 1940 a very

[Begin Page: Page 35]

Fig. 10. Flower color patterns in Nemesia strumosa. For clarity, each

flower is cut along both sides of the tube and is flattened out. Left, the red type with the genotype, C Sp Gr Ro. Center, the splotched modification of red; C sp Gr Ro. Right, the red outline modification; C Sp Grro. Xl6. (From Riley in the Botanical Gazette.)

Fig. 11. Harelip in the mouse. Ventral view of the cranium of a newborn normal (left) and harelip (right) mouse, a, incisor alveolus; b, premaxilla; c, palatine process of maxilla; /, vomer; g, alveolar process of maxilla; h, presphenoid; i, palatine; ;', alisphenoid; k, inner pterygoid process of the alisphenoid; I, basisphenoid. (Courtesy of Dr. S. C. Reed, in the Anatomical Record.)

[Begin Page: Page 36]

Fig. 12. Inherited coat colors in rabbits, (a) normal, CC; (b) chinchilla, c<^''cc^*; (c) Himalayan, c^c^; (d) albino, cc. These genes form a series of multiple alleles. (From Keeler in the Journal of 'Heredity.)

Fig. 13. An adult mouse showing a short-tail mutation. Vertebrae are present at the base of the tail, but not at the tip. (From Dunn, Glueck-sohn-Schoenheimer, and Bryson in the Journal of Heredity.)

[Begin Page: Page 37]

interesting gene that affects the body structure of mice. The dominant gene, Sd, when heterozygous, produces either short or no tails, often shortened or crooked spines, abnormalities of the kidneys, and a generally lowered vitality (Fig. 13). The homozygotes are completely tailless, have spines divided by a cleft into two parts, have no kidneys or external genitalia, and die shortly after birth. This whole complex of characters behaves as a unit.

Some Characters in Human Beings

For several reasons it is far more difficult to study the genetics of human beings than inheritance in plants or in other animals. The technique which is widely used in studying the way the genes of plants and other animals are distributed among individuals, long known as the pedigree culture method, cannot be applied to man because of the social nature of human beings. Also, it is far more difficult to control or at least to analyze the environmental conditions under which human beings develop than those of other organisms, and unless the environment can be eliminated as a variable, our genetic results are always open to criticism. Many of the characters studied in other forms are unit characters in which one gene alone is mainly responsible for the development of a character; in human beings many of

the characters which have been studied appear to be caused by the interaction of a number of different genes and are therefore much more difiicult to analyze. Another complicating factor is that in plants or other animals we can often deal with genes having high penetrance, whereas in human beings many genes seem to have reduced penetrance. Although several hundred characters in human beings have been observed and studied, our knowledge is satisfactory for only a small percentage.

Studies in the heredity of human traits are complicated also by the fact that in some instances two or more genes in different chromosomes may independently produce the same character, or characters that appear the same unless they are carefully studied. In human beings, some of our supposed traits, especially some of the psychological ones, are themselves poorly understood, and until the characters are recognized and distinguished, the genes that are active in their production cannot be identified. Insanity illustrates the last point. Some years ago,

[Begin Page: Page 38]

geneticists were interested in the inheritance of insanity but could make little progress in determining what genes were involved in its production. More recently, the psychologist has informed us that what has usually been termed "insanity" may be any one

of twenty or more different conditions. It is obviously impossible to treat twenty characters some of which may be due to dominant genes and some to recessives, some of which may have high penetrance while others have low, as one character and expect to get a satisfactory genetic analysis.

A number of inherited conditions in man are fairly well known, and it appears that every part of the body and many physiological and psychological processes can be affected by genes or combinations of genes. Sometimes dominant genes are involved and sometimes recessives seem to be responsible. Often the condition is so difficult to analyze or so few cases have been found that on the basis of present knowledge it is impossible to determine the exact nature and number of genes involved even though the available evidence points distinctly to an inherited condition. Some of the more striking or more important discoveries will be cited here, but no attempt will be made to give a complete list of inherited human traits.

Genes have been found which affect the color of the skin. Probably the most familiar is the recessive albino gene which is similar to the albino gene in other animals and completely prevents the formation of pigmentation in the skin, eyes, and hair. Other differences in skin color such as the skin of the black and yellow races in contrast to the white race are usually of interest to most people. In both these situations, multiple genes seem to be involved. Negroes seem to differ from whites by two pairs of genes. In both there is a lack of dominance,

and all four genes interact cumulatively. Thus an individual with the genotype ^4.4. BB would be very dark, whereas an aa 55 individual would be white. Mulattoes with A A Bb or Aa BB combinations would be dark, those with Aa Bb, AA bb, or aa BB would be intermediate, and those with the genes Aa bb or aa Bb would be between an intermediate and a pure white. Other modifying genes might also operate to influence these main types. The gene differences between the white and yellow races also involve several pairs of alleles.

[Begin Page: Page 39]

Negroes with an interesting piebald spotting have been reported by Keeler. Individuals with this dominant gene have normal dark pigmentation in the head, back, hands, and feet except for a white head blaze and a white patch under the chin. Their abdomen, sides, arms, and legs are generally white but are speckled with small patches of normally pigmented skin. The pattern of these individuals is essentially the same as that found in the Dalmatian coach dog, the English rabbit, and Hereford cattle.

Abnormal conditions of skin texture are known. One of the most striking is ichthyosis vulgaris, in which the skin is covered with small, horny flakes or scales. This condition, the result

of a dominant gene, produces the "porcupine men" of the side-show. Cockayne has listed an unusual abnormality, ichthyosis hystri gravior, which occurs only in males. Their entire bodies, except for the face, palms, and soles, are covered with dark brown horny growths which appear after they are two months old. A skin defect that can be quite serious is the inherited absence of sweat glands. Individuals lacking these glands cannot perspire. In warm weather they must go to a cooler region or must remain in water or keep their clothes wet to prevent too great a rise in body temperature. This condition may also affect the skin, nails, teeth, hair, mammary glands, and mucous membranes, and persons with this abnormality may be unable to shed tears. The condition is the result of a recessive gene.

Genes affecting the hair include a dominant gene that produces a white forelock in otherwise dark hair. It was traced for five successive generations in one family. A curious character, the result of a dominant gene, affects the hair in the front of the head in such a way that it falls out when it has grown to five or six inches. When this hair has fallen out new hair comes in, so that affected individuals always have short hair, or bangs, over their foreheads. Other unusual and inherited hair conditions are woolly hair and a condition in which* the embryonic hair continues to grow after birth in such abundance that an individual with this dominant gene can appear in circus sideshows as a "dog-faced" man.

A number of genes affect the axial skeleton, producing abnormalities that may often have a very harmful effect. The gene

for inherited hollow chest reported by Snyder and Curtis pro-

[Begin Page: Page 40]

duces a curious condition that apparently is not harmful in the least. Individuals with this dominant gene have a depression in the chest that looks as if it had been produced by a ball that had been pressed in. A far more harmful gene is the dominant that produces cartilaginous growths on the bones. Another dominant gene affects the bones in such a way that they are easily broken; a person with this gene may have a couple of dozen bone fractures during the course of his life.

Several types of dwarfism, or nanism, are inherited. The ateliotic type or midget, in which the individual is correctly proportioned but much smaller in every way than a normal person, results apparently from the interaction of two dominant genes. The achondroplastic type appears also to be the result of two interacting dominant genes. When both dominants are present, the person has shortened limbs but a normal-sized trunk. Both types of dwarfs may be found in side-shows.

Abnormalities of the fingers and toes are fairly numerous. In Polydactyly, a condition that has been reported a number of times, the individual has extra fingers or toes. A number of

families of the white race have been studied in which this character appears to result from a dominant gene. Negroes with Polydactyly apparently are homozygous for a different gene which is a recessive. Hefner has recorded an interesting case of a dominant gene for Polydactyly which is very variable in its expressivity. In some individuals, the terminal joint of the thumb is long and slender and tends to taper to a point, but the thumb is otherwise normal. Other individuals have thumbs which are long and finger-like and usually bent at a very decided angle toward the index finger. In still others, there may be an extra thumb which is joined to the metacarpal of one or both of these finger-like thumbs while an extra toe is present between the big toe and the normal second toe. This condition appears to be the result of one dominant gene, but it has appeared rather irregularly in several families, indicating probably a gene with reduced penetrance. Other characters affecting the digits are syndactyly, or webbed fingers and toes, which appears to be due to a dominant gene in some families and a recessive in others, brachydactyly, in which a dominant gene results in the absence of the middle phalanx of each finger, causing it to be considerably

[Begin Page: Page 41]

shorter than normal, and minor streblomicrodactyly (Fig. 14) , in which the little fingers are bent. Symphalangism, or the fusion of the phalangeal joints of the digits, is a character caused by

Fig. 14. Crooked little fingers (minor streblomicrodactyly). The abnormality is especially noticeable in side view. (Courtesy of Dr. R. A. Hefner in the Journal of Heredity.)

a dominant gene. Strandskov has reported a case of the inherited absence of thumbnails. This character is probably the result of a dominant gene, although the number of cases is too few for certainty; it is generally accompanied also by a slight abnormality of the nails on some of the other fingers.

[Begin Page: Page 42]

The color of the eyes is greatly affected by certain genes. In albinos, previously mentioned, the eye lacks color entirely (except for the pink color produced by blood in the blood vessels of the eye) because of a homozygous recessive gene. In the presence of the allele of this gene, the eye is colored, but the specific color depends upon the presence of other genes, the exact number of which is not always easy to determine. One pair of alleles seems to produce a basic brown or blue, but these, and especially the brown type, are considerably affected by other genes. The

dominant of this pair, B, produces a purple-black color in the uvea and choroid behind the iris and a brown layer in front of the iris. Because of the latter pigment, the eyes appear brown. In the homozygous recessives, only the first of these two layers is present, and the eyes consequently are not brown and appear blue or gray, depending upon the angle of reflected light, age of the individual, and perhaps modifying genes. The brown type can vary from a very dark brown to a light yellow-brown according to the presence of various modifying genes.

Eye defects are of many kinds. Dominant genes are known which cause ectopis lentis or a congenital displacement of the lens, aniridia, the complete lack of an iris, and glaucoma, a defect in which the normal drainage of the lymph from the eye is blocked and the retina becomes atrophied. Congenital cataract is caused by a dominant gene with incomplete penetrance, for it occasionally fails to appear in individuals possessing the gene. Eye defects produced by recessive genes include a condition in which the optic nerve becomes inflamed and atrophied, and microphthahnus, in which the eyeball is very small and consequently vision is impaired or the affected individual is blind.

In some families a particular defect may be produced by a dominant gene and in others by a recessive. Apparently during the course of evolution, different genes appeared in different individuals and probably at different times, producing the same character or characters which are so nearly alike that they are not separated into different categories. Examples include ex-

treme shortsightedness or high myopia, which is brought about by a recessive gene that produces a globe of unusual length or by a dominant that causes the cornea to be too greatly curved. Farsightedness or hyperopia is a condition in which the globe is so short that the rays are focused behind instead of on the retina.

[Begin Page: Page 43]

In most families it appears to be the result of a dominant gene but in some instances is due to a recessive. In nystagmus the eyeball shows a continuously rolling movement. In some families this is the result of a recessive gene. In others it is caused by a dominant and the character, itself, is somewhat different for the rolling of the eyeball is accompanied by movements of the head. Nystagmus may be caused by environmental conditions as well as by genes. Some injuries to the brain and some brain tumors may cause the same condition as that brought about by the dominant gene. This should caution us against a too hasty judgment as to the inherited or noninherited nature of a certain character.

An interesting ear abnormality reported by Potter is the result of a dominant gene which causes the pinna to be small, deformed, and inverted. Several genes that affect hearing are on record. Deaf-mutes are born totally deaf. This character may be the

result of either one of two recessive genes, d and e, or both. If both dominant alleles are present, the individual is normal, but if a person is homozygous for either d, or e, or both, he is a deaf-mute. Other inherited conditions of deafness are labyrinthine deafness, in w^iich the auditory nerve begins to degenerate at about forty years of age, and otosclerosis, a progressive deafness which begins at about thirty.

A number of inherited tooth defects have been observed. Dominant genes have been identified which cause such abnormalities as absence of the upper incisors, absence of certain incisors and molars, lack of permanent upper canines, lack of two or more wisdom teeth, supernumerary teeth, and defective enamel, resulting in brown teeth. The two center incisors of both jaws are missing in some families as the result of a recessive gene.

In addition to various structural traits, we find that physiological processes and susceptibilities to various diseases may come under gene control. The ability to taste certain substances is an interesting character. Blakeslee and some of his co-workers showed that about seven people out of ten can taste crystals of phenylthiocarbamide. To them, it generally tastes very bitter, although the strength of the taste varies with different people, and to some it appears to be salty. To the other 30 per cent it is tasteless. An examination of a number of families has shown that the ability to taste is due to a certain

dominant gene and that the homozygous recessives detect no taste. Other genes are known that affect the sense of smell and determine whether an individual can detect certain odors.

Other characters that have been reported are hereditary epistaxis or bleeding of the nose. It is due to a dominant gene and may be associated with red spots on the skin and a general susceptibility to colds and nasal infections. An inherited tendency for susceptibility to certain diseases has been noted in many families. A recessive gene for susceptibility to poliomyelitis has been discussed. Recessive genes have also apparently been discovered that produce susceptibility to tuberculosis, scarlet fever, and diphtheria. Studies of inheritance of diseases of this nature are complicated by the fact that a causative infectious agent must be present for the disease to be contracted while other conditions such as general health, insufficient food, and certain conditions under which a person works may contribute to his susceptibility. Considering, however, that every person who is exposed to the disease does not contract it, and considering the pedigrees of a number of people who have contracted it, susceptibility or resistance to these diseases appears to depend in part upon the homozygous condition of a certain recessive gene.

Sugar diabetes or diabetes mellitus is reported to be inherited as a recessive. It may also, however, be caused by syphilis, other diseases, or certain emotional states. That a disease may sometimes be caused solely by environmental factors does not preclude the possibility that it may sometimes be inherited. Actually, this disease may be caused by anything that disturbs the functioning of the islands of Langerhans in the pancreas, resulting in a normal secretion of these structures in a lower amount than in healthy individuals. The cause of the disturbance may be a disease or an emotional condition or a gene.

Certain genes appear to disturb the activity of the thyroid gland, producing such conditions as goiter. Apparently a recessive gene produces alkaptonuria, a condition in which a certain acid, alkapton, is present in the urine, causing it to turn dark. A very rare condition is steatorrhea, a condition in which fat is not digested. It appears to result from one or more pairs of recessives. Another very rare condition, reported by Macklin, is porphyrinuria. Caused by a homozygous recessive, it results

[Begin Page: Page 45]

in the deposition of large quantities of pigment in the tissues, bones, and teeth, and a red color in the urine. A dominant gene seems to be the cause of a general allergic tendency which is

expressed in a great many forms. Some individuals possessing this gene have hay fever, some have eczema, some have hives, some have asthma, and others exhibit still other forms of hypersensitiveness.

The inheritance of psychological traits is, as a rule, more difficult to analyze than the inheritance of physical or of many physiological characters. The most difficult of all is general intelligence, a trait that is not easy to define or to measure. The differences in intelligence between various individuals are not clear-cut and there is a wide range of such differences, with very superior persons at one extreme and very inferior ones at the other and a continuous series of gradations between them. The genetic situation is complicated for apparently a large number of genes is involved in determining intelligence, and they may interact in a very complicated fashion. It is a rather generally accepted view among geneticists that the upper limits of a person's intelligence are determined by his genotype, but how nearly any individual ever reaches his upper limits depends upon a great many factors such as training and other environmental conditions.

Certain grades of insanity are more readily susceptible of genetic analysis partly because they are more readily recognized. Dementia praecox apparently results from the interaction of several recessive genes and is a condition in which a person gradually withdraws into himself and lives in a dream world. The manic-depressive type of insanity is also complex in its inheritance and results from the interaction of several genes,

some of which appear to be dominants. A manic-depressive has alternate periods of great elation and extreme depression. These two types of insanity are hard to analyze genetically, but that they can be identified simplifies the problem considerably. Before they were recognized as distinct conditions and when all types were lumped together under the term "insanity," it was impossible to make a genetic analysis.

Certain other cases of low-grade mentality have been studied sufficiently for at least part of the hereditary cause of the condition to be known. Huntington's chorea is known to be due to a

[Begin Page: Page 46]

dominant gene with almost complete penetrance. Affected individuals show an uncontrollable twitching of head, body, and limbs which develops in the adult and becomes progressively worse. In spinal ataxia, which is due to the interaction of one dominant gene and a homozygous condition at another locus, the afferent nerve tracts become degenerated and the individual loses his sense of equilibrium. Many other inherited conditions of mental disorders might be listed, but those that are mentioned here should probably be sufficient to give a general picture of the need for taking the genotype into account when studying the numerous types of low-grade mentality that are known.

QUESTIONS AND PROBLEMS

1. Breeding results indicate that 103 plants are homozygous for the dominant gene. A, but only 79 of them are A phenotypically. What is the penetrance of that gene?

2. An individual is born with six fingers on one hand, but, because this would make him a curiosity and handicap him socially, the extra finger is removed shortly after birth. Would this affect his genetic constitution? Would it prevent his offspring from having six fingers?

[Begin Page: Page 47]

Chapter 4

REPRODUCTION AND MEIOSIS

It is frequently seen that the same character may be possessed by a number of individuals in different generations of a family, and it is reasonable to assume that these individuals must possess the same gene. One of the important problems of genetics is the way these related individuals came to possess the same gene. It happens that this problem of the distribution of genes is probably the best-understood problem in genetics.

Since the genes are located in the chromosomes, the problem

of gene distribution becomes a problem of chromosome distribution, and since chromosomes are found only in cells the whole problem comes down to a study of how cells are transmitted from one generation to another. Long years of study have shown that at no time during the life of an individual does it receive any cells from its parents except at the moment of reproduction. It is therefore important to understand the various methods by which living organisms reproduce before delving into the manner in which genes are distributed.

The fundamental processes of reproduction are the same in plants and in animals although the details and accessory processes may vary considerably. In the simplest process an entire individual divides into two, but this method is necessarily restricted to the very lowest forms of life. In many organisms a piece of an individual consisting of several cells may develop into a new individual, as in the fragmentation of filamentous algae and of certain coelenterates and flatworms, in the formation of buds in Hydra and some sponges, in the gemmules of certain sponges and the gemmae of liverworts, and in various types of vegetative reproduction in the higher plants. None of these methods involves the union of any cells; they are examples of asexual reproduction.

In contrast to asexual reproduction is sexual reproduction, which involves typically the union of two cells known as germ

[Begin Page: Page 48]

cells or gametes. Some lower forms reproduce sexually by the fusion of identical gametes, but a differentiation of the gametes into male and female is the rule in the higher groups.

Reproduction and Life Cycles in Higher Animals

Sexual reproduction in the higher animals is generally brought about by the union of two unlike gametes, each of which is contributed by a different individual. One gamete, the egg, is large and nonmotile, is produced by the female, is usually spherical, and contains a nucleus and cytoplasm. In the cytoplasm is found the food or yolk. The male gamete, which is much smaller than the egg, is known as the spermatozoon or simply the sperm. It usually consists of three parts: the head, middle piece, and tail. The head is essentially a nucleus surrounded by a very thin layer of cytoplasm and is generally spherical or elliptical in shape. The middle piece is much smaller than the head and, at least in some animals, contains a centrosome. The tail is very long, extremely delicate, and is a flagellum which propels the sperm from place to place. The union of an egg and a sperm is called fertilization. This process results in a cell, the zygote, which will develop into a new adult

individual of the same species as the parents.

It was stated previously that every cell of the body, with the exception of the germ cells, contained the same number of chromosomes. In man, the number in the somatic cells is 48 chromosomes or 24 pairs. If the germ cells were produced by typical mitotic divisions and, therefore, if they had the same number of chromosomes as the body cells, the number of chromosomes of a species would double each generation. Thus it would not be long before the chromosome number of all organisms would be in the thousands and even millions. Actually, however, this does not happen for, with the development of sexual reproduction, a modification of the ordinary mitotic process has developed which, in animals, produces gametes with half the number of chromosomes as in the body cells of the parents. Thus in each generation of human beings the body cells have 48 chromosomes and the gametes have 24. In discussing animals in general, without reference to any particular species, it can be said that the body cells contain 2n chromosomes and the germ cells contain n, where n stands for a definite number. This num-

[Begin Page: Page 49]

ber happens to be 24 in man. The 2n or somatic number is called the diploid (Greek, diploos, twofold, double; Latin, du-

plex) number whereas the n or gametic number is generally referred to as the haploid (Greek, haploos, single; Latin, simplex) and occasionally as the monoploid (Greek, monos, alone, only) number.

The haploid number in the gametes is also called the reduced number and is brought about by two successive mitoses which differ from ordinary mitotic divisions in several important details. These peculiar mitoses do not occur in all parts of the body but only in the ovary and testis. To differentiate them from somatic cell divisions, these two divisions are called the reduction divisions, meiosis, or, because they occur during the maturation or differentiation of the germ cells, the maturation divisions. There are always two meiotic divisions, and consequently each cell that divides by meiosis produces four cells.

In the male animal, a number of cells in the testis become set apart and generally become larger than the others. They are the primary spermatocytes and they undergo the first meiotic division, by which each forms two secondary spermatocytes. The second meiotic division immediately follows, with the result that four spermatids have been produced from each original primary spermatocyte. These spermatids do not divide further, but usually change their shape by elongating and by developing a tail. Each becomes a mature spermatozoon.

In the ovary, when the eggs are about to form, certain cells become very large. These primary oocytes divide to form two cells but they are not alike. One is large and contains all the

stored food; the other is no more than a nucleus with some cytoplasm around it. This small cell is the first polar body and remains attached to the large cell which is the secondary oocyte. The secondary oocyte and frequently the first polar body, also, then undergo the second meiotic division. The secondary oocyte farms a large functional egg and a small, nonfunctional secondary polar body, whereas the first polar body, if it divides, forms two polar bodies. Thus each primary oocyte forms either three or four cells, but only one of them is functional.

In the higher animals, all the somatic cells are diploid. The eggs and sperm of the animals possess the haploid number of chromosomes and unite to produce new individuals which like

[Begin Page: Page 50]

the parents have the diploid number. A succession of events, starting with one stage of life and including all the steps that occur until a new individual at the same stage as the first is found, is called the life cycle of the organism. The life cycle of most animals, including the vertebrates, is very simple.

Reproduction and Life Cycles in Higher Plants

In plants above the Thallophytes and, indeed, in many algae

and fungi, reproduction is complicated by a more involved life cycle than is generally found in the Animal Kingdom. Although we frequently think that we can recognize the body of a certain kind of plant, few of us except botanists recognize that in all these higher plants the complete life cycle includes two different plant bodies. In the fern plant, for example, we are all familiar with the often large, leafy structure that bears typical "fern leaves." These leaves bear minute spores in clusters on their under side. When these spores germinate, they do not produce typical fern plants but small, flat, green bodies, perhaps half an inch long or less, which lie close to the ground. These bodies are fern plants just as much as the more familiar types and they bear the gametes. When the gametes unite, a zygote is produced which develops into the familiar type of fern plant. Thus the complete life cycle of a fern includes two bodies: the large body on which these spores are found, the sporophyte, and the small body that bears the gametes, the gametophyte.

The meiotic divisions in the fern occur not in the formation of the gametes, as in animals, but in the development of the spores. The sporophyte plant has 2n chromosomes and produces haploid spores. They germinate and by a series of regular mitotic divisions produce the haploid gametophyte, which in turn produces haploid gametes. They unite to form a diploid zygote, which, in turn, develops into the diploid sporophyte body. This alternate production of sporophyte and gametophyte bodies is called alternation of generations.

The existence of two generations in the life cycle of the higher plants can best be grasped from such a plant as the fern, where each generation is throughout most of its life a separate and independent structure. In the seed plant the gametophyte is reduced in size and complexity to only a few cells.

[Begin Page: Page 51]

In the angiosperms, the sporophyte, commonly regarded as the plant itself, bears two kinds of spores which in turn produce two kinds of gametophytes. The male spores or microspores are formed in the anthers of the flower. Cells towards the inside of the anthers enlarge and become microspore mother cells or microsporocytes. They undergo the usual two meiotic divisions, and each forms four microspores. A microspore is a round cell with one nucleus and, as it develops, it secretes about itself a thick wall which is usually yellow and highly sculptured in such a characteristic way that the species of plants can be identified from the ridges and furrows of the microspore walls. A microspore is a one-nucleate pollen grain; but soon after it is formed, the nucleus and sometimes also the cytoplasm divide into a tube nucleus or tube cell and a generative nucleus or cell. This two-nucleate or two-celled pollen grain is a microgametophyte.

The female spores form from megaspore mother cells or mega-

sporocytes, located in the ovules. The female organ is the pistil of the flower. It is enlarged at the base into an ovary which contains one or more ovules, each of which can develop into a seed. Each ovule contains only one megasporocyte, and it divides, by meiosis to form a row of four cells, each of which is a potential megaspore. Three of these cells degenerate and the fourth enlarges to form a large functional megaspore or young embryo sac. The nucleus divides by ordinary mitosis to form two, four, and finally eight nuclei within the one embryo sac. Three nuclei collect at each end and two in the center, and cell walls are formed about the three at each end. One of the cells at one end of the embryo sac is the female gamete or egg. The embryo sac at this stage is mature and is the female gametophyte or megagametophyte.

When the two-celled pollen grain is mature, it is liberated from the anther and blows or is carried by insects to the end of the pistil, where it adheres. The wall of the pollen grain bursts, and the protoplasm grows out as a pollen tube which grows down through the tissues of the pistil until it enters the ovary. The pollen tube is a later stage of the microgametophyte. The tube nucleus precedes and the generative nucleus follows farther behind in the tube (Fig. 15). As the tube approaches the ovule^ the generative nucleus divides by mitosis to form two sperm nuclei or male gametes. The tube then passes through the micro-

[Begin Page: Page 52]

pyle, a small hole in the ovule, and enters the embryo sac. The tube nucleus disintegrates, while one sperm nucleus fertilizes the egg to form the zygote and the other unites with the two nuclei at the center of the embryo sac, the polar nuclei, to form endosperm, sl tissue in which food is stored for the developing embryo. The zygote is the first stage of the new sporophyte.

Higher plants have a much more complicated life cycle than animals because two generations are necessary to complete the entire cycle.

Meiosis

The basis of the difference between the two meiotic divisions and any two successive somatic mitoses is to be found at the beginning of the prophase of the first of the two meiotic divisions. It has been pointed out that the chromosome in the resting stage is either a single structure or is composed of two chromonemata which are in such an intimate re-

lationship that they behave as a single structure. By the beginning of prophase in an ordinary somatic mitosis, either the single chromonema has doubled or the two chromonemata of each chromosome have become sufficiently separated that the chromosomes are definitely double structures. At the prophase of the first meiotic mitosis, however, the chromosome is still effectively single, just as it was in the resting stage. This is a fundamental difference between mitosis and the first meiotic division.

Another important difference is that shortly after the chromosomes appear, they begin to lie alongside one another in pairs. Each chromosome pairs with the chromosome which is identical with it in size and shape — in other words, with its homologue. In meiosis, it is not until ajter the chromosomes have paired that each chromosome becomes a double structure. After each chromosome has become double, a chromatid from one homo-

FiG. 15. Pollen tube showing two sperm nuclei. (Courtesy of Dr. George H. Conant.)

[Begin Page: Page 53]

logue may break and the broken pieces join up with broken pieces of a chromatid of the homologous chromosome. All these processes occur in the first prophase, and consequently this stage is of longer duration than the prophase of ordinary mitotic divisions. Cytologists have found it convenient to subdivide the first meiotic prophase into five substages.

First Projphase. At the beginning of the first prophase, the chromosomes are present in the diploid number just as in a mitotic prophase, but they are single throughout and not double. This stage of the first meiotic prophase is known as leptotene. In leptotene the chromosomes are very long, thin threads probably corresponding to the chromonemata of the anaphase chromosomes of the preceding mitotic division. They have a more granular appearance than the chromosomes in prophase of a somatic mitosis, and they often appear as loosely strung strings of beads of unequal size (Fig. 16). They are coiled loosely in relic coils of the previous division. Other morphological differentiations are not usually noticeable except that the centromere is frequently stained more lightly and may at this stage appear thicker than the rest of the chromosome.

Shortly after the chromosomes have appeared, they begin to pair up, each with its homologue. This pairing or synapsis, which occurs during zygotene and continues until all the chromosomes are completely paired, is very precise, for each part of a

chromosome will lie exactly alongside the corresponding part of its partner. This is true to such an extent that if pieces of one chromosome are broken away or inverted, that chromosome and its homologue will twist about so as to bring corresponding parts together (Fig. 17). In most higher plants and in some animals in which the chromosomes lie at random in the nucleus throughout leptotene and zygotene, the homologues may begin to pair at any place, but sometimes they do so at the centromere or at the ends. In many animals and in some higher plants the chromosomes may have the orientation of the preceding telophase so that one or sometimes both ends are pointed towards one region of the nucleus. In organisms with definitely oriented or polarized chromosomes, pairing usually begins at the ends nearest the nuclear membrane and continues along the chromosomes until they have completely paired. When the chromo-

[Begin Page: Page 54]

a

g

h

j k l

Fig. 16. Meiosis in plants: (a) leptotene; (6) zygotene; (c) pachytene; (o?) diplotene; (e) diakinesis; (/) first metaphase; {g) first anaphase; {h) first telophase; (i) second prophase; (;') second metaphase; (/c) second anaphase; (0 second telophase. Diagrammatic.

[Begin Page: Page 55]

somes are polarized they are sometimes said to be in the bouquet stage.

Zygotene is followed by pachytene, a stage characterized by several important featm'es. The two homologous chromosomes which had paired in zygotene now twist about one another in

Fig. 17. Chromosome pairing in which one of the pairing homologues has a deleted or an inverted segment: (a) a terminal deletion; (b) an intercalaiy deletion; (c) an inversion. (Courtesy of Dr. B. McClintock in Research Bulletin of the University of Missouri Agricultural Experiment Station.)

what are called relational coils (Fig. 18), and each soon becomes a double structure. It is immaterial for our purpose whether this is because two chromatids in very close contact and constituting one chromosome become so separated now as to be visible as a double structure or whether one original thread w^as present which now forms another thread like it and alongside it. It is often stated that at this time "the chromosomes split longitudinally," but it is more likely either that two intimate chromonemata separate or that one thread regenerates another. After this doubling occurs, the two chromatids of one chro-

[Begin Page: Page 56]

mosome are still twisted about the two chromatids of the homologous chromosome in relational coiling, but this is further complicated by a coiling of the two chromatids of each chromosome around each other. Thus two threads (chromatids) which are coiled about each other are coiled relationally about two other

Fig. 18. One bivalent in stages of the first meiotic division. Top, zygotene or early pachytene before the chromosomes have become double. Center, pachytene with each chromosome consisting of two chromatids. Bottom, diplotene showing chiasmata.

threads (chromatids) which are coiled about each other. It can

be seen from this that the chromosomes are under considerable stram. Before the chromosomes become double, there is an attraction of an unknown nature which causes them to remain paired. Once the chromosomes are doubled this attraction ceases and is translated into an attraction between the chromatids of each pair. AVhen the attraction between chromosomes lapses, one pair of chromatids begins to repel the other pair, increasing the strain. The result will be that at one or more places one of the four threads will break, and it will not necessarily be the same thread that breaks at any two places.

[Begin Page: Page 57]

When one chromatid breaks at a certain place, one of the two chromatids of the other chromosome also breaks at exactly the same place. The broken ends then uncoil. The broken end of one chromatid in some way seems to come into contact with the broken end of the other broken chromatid and they fuse. This fusion largely eliminates relational coiling. If one such break, followed by a fusion, occurs, one chromatid of each chromosome remains unchanged but the other two chromatids are new and are composed in part of one original chromatid and in part of the other. Since the break is at the same place in each of the two chromatids, the new chromatids are exactly the same as the old ones in size and appearance ; but the new arrangement

of segments of chromatids has a very important effect on the transmission of groups of genes, resulting in what is known genetically as crossing over. Since more than one break-and-fusion usually occurs in a pair of homologous chromosomes, and since they may involve any chromatid of either pair at any one place, the results in terms of the original nature of the four threads can be quite complicated.

The breaks and exchanges of partners produce cross-shaped figures in the paired chromosomes when viewed under the microscope. They are best observed in the next stage of the first meiotic prophase, diplotene. During diplotene, the repulsion between the pairs of chromatids is very strong, and the two pairs now tend to separate from one another. They cannot do so completely, however, because at various places one chromatid from one chromosome is attached to a piece of one chromatid from the other chromosome. The parts that are not joined separate as widely as they can. If one break had occurred in pachytene, the two homologous chromosomes would present a cross-shaped figure, and the length of the arms of the cross would depend upon the original position of the break. If more than one break had occurred, the chromosomes would open out into loops. The regions where they are tied together as the result of the previous breaks are known as chiasmata.

The two homologous chromosomes which have paired at zygotene are known as a bivalent. When four threads are present as the result of the doubling of each chromosome, the configuration is known as a tetrad. The tetrad nature of a bivalent is not

[Begin Page: Page 58]

so easily observed at pachytene because the threads lie close together, but at diplotene as the result of the repulsion and consequent opening out of the threads, the four-strand nature is easily seen. The diplotene chromatids are long and thin but, as diplotene progresses, they contract greatly and become much thicker. This contraction is due to the coiling or spiralization of the long, thin threads that were present originally. At the same time, they rotate in such a way that in a bivalent with several chiasmata the successive loops lie at right angles to one another, whereas if only one chiasma is present, the arms rotate through 180°. As at mitosis, a matrix which stains very heavily begins to collect around the threads so that the internal structure of the chromosome is not easily visible late in this stage. The repulsion which starts with the beginning of diplotene continues and is often strong enough to cause the chromatids to slide along one another so that the chiasmata appear to move towards the ends. This terminalization begins towards the end of diplotene and may continue through the next stage and up to metaphase. Chiasmata may terminalize completely or only partially. Terminalization is, in general, greatest in small chromosomes and least in large ones, although the degree of terminalization also seems partly a characteristic of certain species.

The chromosomes pass gradually from diplotene to the last stage of the prophase, diakinesis. At this stage, the bivalents are very short and thick and are quite deeply stained, and the two chromatids of each chromosome are close to one another, with the result that the identity of the individual chromatids is usually lost except possibly at the ends when the chiasmata are not completely terminalized. During this stage, the spiralization of the chromatids may continue, causing them to become somewhat shorter than at the beginning of diakinesis, and terminalization may also continue if it had not been completed in diplotene. The bivalents tend to repel each other during diakinesis. They move to the periphery of the nucleus just inside the nuclear membrane and are frequently arranged so that each is as far away from every other one as it can get, although this last feature seems more noticeable in small than in large nuclei. The nucleolus disappears during diakinesis, and this stage is terminated by the disappearance of the nuclear membrane.

[Begin Page: Page 59]

First Prometaphase. When the nuclear membrane disappears, a spindle forms and the bivalents move towards the equator. The chromosomes are even shorter and thicker than they were during prophase. The chromatids are not usually visible under

ordinary methods of staining but can be made to appear by special techniques.

First Metaphase. AVhen the bivalents reach the equator they arrange themselves on it. They are especially clear in plants with few chromosomes (Figs. 19 and 20). If the plant has both large and small chromosomes, the small ones are usually towards the center (Fig. 21). There are several outstanding differences between metaphase of the first meiotic division and a metaphase of a somatic mitosis. In a somatic division, the metaphase chromosomes are placed so that their centromeres lie on the equator. In the first meiotic metaphase, the centromeres could not lie on the equator unless the bivalent lay on its side. The bivalents are oriented so that the centromeres are towards the poles and the chiasmata in the equatorial plane.

The way any bivalent is oriented is purely a matter of chance. That is, the centromere of the chromosome which came from the male parent may point to either pole, and obviously the centromere from the original female parent will point in the direction of the opposite pole. Not only is any one pair of chromosomes oriented at random with respect to the poles but each bivalent is also arranged entirely independently of any other.

First Anaphase. When the bivalents have become arranged on the spindle, they begin to pull apart. This separation is apparently the result of a strong repulsion between the opposite centromeres, wiiich drag the rest of the chromatids after them.

When the chiasmata are terminalized, the chromosomes break apart easily as the centromeres move towards the pole; when nonterminalized, the chiasmata slip along towards the ends until the chromosomes have pulled apart. The two sister chromatids are still in contact at the centromere, but the double nature of each chromosome, often completely obscured at metaphase, is now very evident. The anaphase chromosomes are much shorter and thicker than the anaphase chromosomes in a somatic mitosis of the same plant and would hardly be recognized as belonging to the same organism.

[Begin Page: Page 60]

Fig 19 Stages in the development of the pollen grams m T'«descanlm Jludola- (aTfirst metaphase; (6) first anaphase; (c) second anaphase W r^etanhle ot the microspore or first postmeiot.c division; («) early lllpZt and (/) lite anaphaL of the microspore division. Camera lueida drawings.

[Begin Page: Page 61]

[Begin Page: Page 62]

First Telophase. After the anaphase chromosomes have reached their respective poles, they frequently become very long and they may largely uncoil as in mitosis. A new nuclear membrane may form around each group of chromosomes, constituting

Fig. 21, Metaphase and anaphase of the first meiotic division in two plants with chromosomes of very different size: (a) metaphase of Gasteria nigricans; (b) anaphase of Gasteria laetepunctata ; (c) metaphase of Capsella (Bursa) rubella; (d) anaphase of Capsella grandifiora. In Gasteria there are four pairs of very large and three pairs of much smaller chromosomes. In these species of Capsella there are eight pairs of very small chromosomes, (a) and (b) X930; (c) and (d) circa X2000. Camera lucida drawings.

two daughter nuclei, a nucleolus may appear in each nucleus, and a new cell wall may now form, dividing the original cell into two. However, sometimes the first telophase is apparently

dispensed with, and the chromosomes may pass unchanged from the first anaphase into the second prometaphase.

Interkinesis. After the first telophase, the daughter nuclei sometimes go into a typical resting stage just as they do after

[Begin Page: Page 63]

a somatic mitosis. This stage between the end of the first telophase and the beginning of the second prophase is known as interkinesis. It is usually short, and may be entirely absent. Sometimes the chromosomes seem to go into a partial but not complete resting stage between the first and the second prophase.

Second Prophase. If an interkinesis follows the first telophase it is in turn followed by the prophase of the second meiotic division. In the second prophase the chromosomes appear as double structures, the result of the "split" or separation of the chromatids which took place at pachytene or one whole cell division previous to their appearance here. The chromatids of each chromosome are held together by the centromere, but the arms repel one another instead of lying in close approximation as in a somatic mitosis. This arrangement gives the chromosomes a very different appearance from the prophase chromosomes of a somatic mitosis for in the second meiotic division

they are X-shaped figures whereas in a somatic mitosis they are two parallel threads. In a somatic mitosis there are 2n prophase chromosomes, but in the second meiotic prophase the chromosomes are present in only the haploid number. During the second prophase, the nucleoli, if they appeared during the first telophase, disappear again, and finally the nuclear membranes disappear.

Second Prometaphase. When the two nuclear membranes break down, two new spindles are formed in the position of the former nuclei, and the chrom.osomes of each nucleus move on to the equators of their respective spindles. If a new cell wall formed at first telophase, each spindle is in a separate cell, but if the wall did not form, as is normal in many organisms, both spindles are in one cell. The two spindles may lie approximately parallel and alongside one another, as in spermatogenesis in animals or the formation of the microspore in plants. Then the four cells which form subsequently are arranged in the form of a tetrahedron. In megasporogenesis in plants, however, the spindles are oriented in the same direction and lie in the same plane; the resulting four cells are in a linear row.

Second Metaphase. As in somatic mitosis, the chromosomes are lined up with the centromeres on the equator. The repulsion of the arms found at second prophase is now completely or partially overcome, and the two chromatids of each chromosome

often lie close together as in a somatic mitosis, although they sometimes diverge. Where there is no interphase, the spindle of the first division seems to break up into two spindles just as the anaphase chromosomes of the first division have reached the poles, and the two groups of anaphase chromosomes move immediately on to the equatorial plates of the new spindles and become the second metaphase chromosomes. They usually elongate in the process.

Second Anaphase. Second anaphase begins when the daughter centromeres pull apart towards the opposite poles. The chromosomes of second anaphase are not the short, thick bodies of the first meiotic division but are much more like the anaphase chromosomes of a somatic mitosis.

Second Telophase. When the anaphase chromosomes reach the poles, new nuclei form in the usual manner. The chromosomes lengthen and almost completely uncoil, and nuclear membranes and nucleoli appear. Cell walls usually divide these two cells into four although occasionally no walls form, as in megasporogenesis of the lily. In organisms in which cell walls do form, if a wall did not form during the first telophase, the one cell with four nuclei now becomes divided into four cells. -

If an animal has 16 somatic chromosomes, at leptotene there would be 16 chromosomes and therefore 16 centromeres. After pairing and "splitting" of the chromosomes there would be 32 chromatids; but since the centromeres either do not divide or divide but remain with the daughter centromeres in very intimate contact during the first division, only 16 effective centromeres would still be present. At first anaphase, 8 effective centromeres and therefore 8 chromosomes would pass to each pole. The fact that each chromosome was composed of 2 chromatids would not make it more than one chromosome for, as long as the centromeres are intact or together, the chromosomes behave as a unit irrespective of the number of chromatids of which they are composed. Therefore, at first anaphase, 8 chromosomes go to each pole. In the second division, the daughter centromeres separate so that each chromosome now becomes two separate units. As a result of this separation of the centromeres, 8 chromosomes go to each pole at the second anaphase.

[Begin Page: Page 65]

At the beginning of meiosis the primary spermatocytes and primary oocytes of the above animal have 16 chromosomes. At the end of the first division, the secondary spermatocytes and

the secondary oocytes have only 8. Since the nmiiber is reduced half, this division is often referred to as the reduction division. The reduction division reduces the number of chromosomes and centromeres. In the second meiotic division, the chromatids of each chromosome separate from one another. Because of the "split" of the centromeres, there is no further reduction of the number of centromeres or of chromosomes in each daughter cell. It is not, therefore, a reductional division, but is often called the equation division because of the equal separation of sister chromatids to the daughter nuclei.

Usually the first division is reductional and the second equational for the number of effective centromeres and for the 7iumber of chromosomes. Because of breaks in the chromatids and the formation of new combinations of segments of chromatids at pachytene which result if chiasmata occur at diplotene, reduction is true only in a quantitative sense and is not true qualitatively for the whole of all the chromatids. If there were no exchanges of chromatids and no chiasmata (assuming that the chromosomes were still paired), the entire homologues would separate reductionally in a cjualitative sense at first anaphase and equationally at second anaphase. Normally, however, one or more chiasmata are formed. In a chromosome having one long and one short arm, let us assume that one chiasma can form in the long arm but that the other arm is too short for a chiasma. The result is a chromosome passing to one pole at first anaphase which is composed of one normal chromatid plus a sister chromatid which is normal for the short arm and the part of the long arm nearest the centromere (the proximal part) but has a

distal piece of a homologous chromatid. Similarly, the chromosome passing to the other pole is normal except for the corresponding distal segment. As a result, the first anaphase is reductional for the centromere, short arm, and proximal piece of the long arm, but is equational for the distal end of the long arm of one chromatid of each chromosome. Correspondingly, the second division is equational for the short arm and proximal part of the long arm but reductional for the distal portions. If more than one chiasma is present, the chromatids are more complex.

[Begin Page: Page 66]

Plant morphologists have frequently called the first meiotic division the heterotypic division, or division different from a typical mitosis, and the second meiotic division the homeotypic division, or division like a typical mitosis. In number of chromosomes involved and in the position of the arms of the "split" chromosome in prophase, however, this second meiotic division differs from a somatic division.

Vegetative Reproduction

In higher plants various vegetative methods of reproduction may be found by which a new plant may arise from a piece of one of the vegetative organs of another. Such asexual methods

of reproduction may be the only usual methods in some plants, and in many plants they may be of great importance from an agricultural viewpoint.

If plants reproduce by a vegetative method, whether from roots, stems, or leaves, all the new plants will be exactly like one another and like the parental plant. A group of plants produced vegetatively from one original plant is called a clone. All the plants of a given clone are alike.

Hermaphrodites

In many animals and some higher plants, each individual is either male or female; but in some animals and most higher plants, both sexes are represented in each individual. Organisms in which one individual contains both male and female sex organs are known as hermaphrodites. In some hermaphroditic animals, like Hydra, the sperm of one animal will fertilize the eggs of the same animal; but in the earthworm, the eggs must be fertilized by sperm from a different animal. When an hermaphroditic animal or plant produces a new individual by the union of egg and sperm from the same parent, such an organism is said to be self -fertilized; but when the gametes are from different individuals, such an organism is cross-fertilized. Many seed plants have elaborate mechanisms to ensure that they will be cross-fertilized, peas and others are regularly self-fertiUzed, and still others may produce seeds by either self- or cross-fertilization.

[Begin Page: Page 67]

Questions and Problems 67

Parthenogenesis

Eggs normally require fertilization in order to develop into mature organisms, but the eggs of some plants and animals may develop without fertilization. The development of an unfertilized egg is known as parthenogenesis. Although this occurs normally in the production of certain insects such as male bees and the parasitic wasp Habrobracon, it can be induced in some eggs by treating them with certain chemicals or other abnormal environmental conditions.

QUESTIONS AND PROBLEMS

1. Consult books on general zoology and suggest some animals that have a more complicated life cycle than the vertebrates. What are the chromosome numbers of the various stages of some of these other life cycles ?

2. Diagram the life cycle in Ulothrix, Fucus, Nemahon, Polysiphonia, black stem rust of wheat, and other lower plants. Consult textbooks on general botany or on the morphology of the Thallophytes.

3. Discuss the relative importance of the sporophyte and gametophyte in various divisions of the Plant Kingdom.

4. The number of chromosomes in the root tip cells of maize is 20. What is the number in the following cells or tissues: (a) microsporocyte ;

(b) tube nucleus; (c) nucellus; (d) antipodal cells; (e) cells of anther wall; (/) style; (g) embryo sac mother cell; (h) megaspore; (i) palisade cells of leaf; (;') endosperm?

5. Assume that a plant has two long and two short chromosomes. Diagram cells of that plant in (a) metaphase and anaphase of a somatic mitosis, (6) metaphase and anaphase of the first meiotic division, and

(c) metaphase and anaphase of the second meiotic division.

6. Explain what is meant by relational coiling and relic coiling.

7. What is meant by Darlington's "Precocity Theory"?

8. If several chiasmata form in one arm of a chromosome and if they all terminahze, why do they all terminalize to the free end of that arm? Why do not some terminalize to the end of the other arm? Would they still all terminalize to the end of that arm if no chiasmata formed in the other arm?

9. Show by a diagram why a chiasma holds two chromosomes together at diplotene. Use colored crayons to differentiate the two homologues or, still better, use colored modeling clay.

[Begin Page: Page 68]

Chapter 5

SPECIAL CHROMOSOMES AND SEX INHERITANCE

Sex and the Sex Chromosome

For most animals and a few plants, there is one outstanding and regular exception to the statement that all the chromosomes of a normal, diploid organism are in pairs and that each chromosome has a mate which is an exact duplicate of it in morphology and in the loci of which it is composed. In all individuals of Drosophila melanog aster, the second, third, and fourth chromosomes are present in pairs, and the first chromosome, rod-shaped and of medium length, is paired in the female. In the male, however, only one of these medium-sized rod-shaped chromosomes is present, and another chromosome, which is longer, J-shaped, and absent in a nor-

1-^^ ^^ ^^ mal female, is also present. It

m\ is customary to refer to the rod-

shaped chromosome as the X

Fig. 22. Chromosomes in a fe- chromosome and the J-shaped male-determining {lejt) and a ^^^ ^g ^j^^ y chromosome. At

male-determining sperm of Dro- • • • ,1 a ±^ -v

,., , , -Pk- ^ meiosis m the male, the X

sophila melanogaster. Diagram-

matic. (After Bridges in Genetics.) and Y chromosomes separate

so that all sperm carry one X

or one Y in addition to one chromosome from each of the three pairs (Fig. 22). Since females have two X chromosomes, all eggs have one X chromosome in addition to one member of each of the other pairs. The X and Y chromosomes are therefore differentials in the determination of sex. When an X-bearing sperm unites with an egg, the resulting individual has two X chromosomes and is a female. Similarly, a male is produced by the union of a Y-bearing sperm and an egg, since it has one X and one Y chromosome (Table 1).

jk Jk

The chromosome mechanism that explains sex in Drosophila is not universal as to details, and yet the fundamentals are the same in all organisms in which the sexes are separate. This type, in which the male has both an X and a Y chromosome while the female is XX, is the more general condition, although the Y chromosome is not J-shaped in all organisms, nor is it always larger than the X chromosome. In human beings, for example, the Y is a very short chromosome and is considerably smaller than the X. In human beings, 48 somatic chromosomes are present; half the sperm have one X chromosome and 23 others

TABLE 1

Sets of Autosomes (A) and Sex Chromosomes (X, Y, Z, axd W) in Females

AND Males, in Their Gametes, and in Their Offspring in Diploid

Organisms Having the XY and in Those with the ZW Type of

Sex Chromosomes

and are female-determining, and half have a Y chromosome and 23 others and are therefore male-determining. Since the sex chro-

mosomes are so important in determining sex, the sex of a child is determined at the time of fertilization. Because of the nature of the sex-determining mechanism, theoretically half of all children born should be boys and half girls. Actually, the ratio is about 105 boys to 100 girls, a slight deviation from theoretical expectations hard to account for. It has been supposed that the male-determining sperm move just slightly faster than the other type, but experimental proof is lacking.

In such organisms as grasshoppers and certain bugs, the female is XX and the male is XO. In other words, there is no Y chromosome, and the male thus has one chromosome less than the female. This situation, sometimes referred to as the Protenor type, was the first one discovered; the unpaired chromosome in the male was called the "accessory" chromosome before its function was realized. This type is similar to the Drosophila type except that

[Begin Page: Page 70]

half the sperm have neither an X nor a Y chromosome and are male-determining.

Autosomes

In all organisms the eggs and sperm carry one member of

each of the other pairs of chromosomes in addition to the X or Y chromosome. These other chromosomes are known as the autosomes. In Drosophila melanogaster there are 6 autosomes in the somatic cells of both the male and female and 3 in each gamete; in man, there are 46 autosomes in each body cell and 23 in each gamete.

Heterogametic Females

In moths, butterflies, birds, and some fishes, the situation as regards the sex chromosomes is the opposite from that in Drosophila and man. In these organisms, the female is the heterogametic sex. This type is generally referred to as the Abraxas or bird type, and the sex chromosomes in the female are usually designated Z and W whereas the male is ZZ. Inheritance of sex in these organisms is shown in Table 1.

The Y Chromosome

Apart from its frequent difference in shape, the Y chromosome differs in one marked respect from the X chromosome and from the autosomes. It generally contains at most just a few genes. Often no genes at all have been discovered in the Y chromosome, and even where genes have been found they are frequently not alleles of genes in the X chromosome. In the Y chromosome of Drosophila melanogaster genes that have been discovered are a gene for long bristles, which is an allele of the gene "bobbed" in the X chromosome, and two genes for male fertility which appear

to have no corresponding allele in the X chromosome. The presence of the allele of bobbed indicates that there is one small segment of the X chromosome which is represented by a homologous segment in the Y chromosome. The remaining parts of the X chromosome have no counterpart in the Y chromosome, and almost all the Y chromosome is completely nonhomologous with the X chromosome. Apart from the few genes, the Y chromosome of Drosophila appears to be made up of inert material usually

[Begin Page: Page 71]

called heterochroinatin to distinguish it from the active or euchromatic regions. These two types of chromatin stain somewhat differently during mitosis. Although most of the Y chromosome is inert, heterochromatic material is not confined to the Y chromosome, for about one-third of the X chromosome nearest the centromere and small regions of the autosomes on either side of their centromeres appear to be heterochromatic.

As in Drosophila, there is an homologous segment in both the X and Y chromosomes of human beings, but this segment is small in comparison with the nonhomologous regions and only a few genes have been found in it. A number of loci are found in the X chromosome which are not represented in the Y chromosome, and several genes are known in the part of the Y chromo-

some not represented in the X chromosome. Some of the characters produced by genes on. the X and Y chromosomes in man have been mentioned in Chapter 3.

Meiosis in the Sex Chromosomes

The meiotic behavior of the chromosomes in individuals with two X chromosomes, such as the females of many species, or with two Z chromosomes, as the males of birds and a few other animals, is exactly like the meiotic behavior of the autosomes. This is to be expected since the two X chromosomes are homologous throughout their length. Thus they pair at zygotene, exchange segments, and form chiasmata in exactly the same way as autosomes. In the heterogametic sex, however, the behavior depends upon the presence or absence of the Y chromosome and, if a Y chromosome is present, upon the extent of homology between it and the X chromosome. In organisms of the XO type naturally there is no chromosome with which the X chromosome can pair (Fig. 23). It will usually pass intact to one or the other pole at the first meiotic anaphase and divide equationally at the second. If it does so, it goes to the pole either before or after but never at the same time as the autosomes. Sometimes the univalent X chromosome divides equationally at the first division and reductionally at the second. In either case only two of the four resulting cells contains an X chromosome. In the XY type in most organisms, pairing may occur between the X and Y chromosomes provided they have a segment in common, but pairing is always between homologous

[Begin Page: Page 72]

Fig. 23. Meiosis in a male squash bug, Anasa tristis. The sex chromosomes in the female sex are XX and in the male XO. (a) Late prophase of division of primary spermatocyte showing nine bivalents, one unpaired X chromosome, and two small univalents which have not paired but will pair as the spindle forms, (b) First meiotic metaphase in polar view; the paired small chromosomes are in the center and the large X chromosome is toward the outside of the principal ring, (c) First anaphase ; the small chromosomes separate in advance of the others; the X chromosome divides equationally at the first division and is shown separating to the poles slightly later than the autosomes (which are dividing reductionally) . (d) First telophase; the X chromosomes appear to be outside the principal ring formed by the others, (e) Second metaphase in polar view. (/) Second anaphase ; the X chromosome passes undivided to one pole but later than the others; the equational splitting at the first division and the passing reductionally at the second spermatocyte division have been termed postheterokinesis. (g) Second telophase; one cell has nine large and one small autosomes, and the other has nine large and one small autosomes and one X chromosome. Xl^25. Camera lucida drawing.

[Begin Page: Page 73]

parts only. If the homologous parts are not too short, chiasmata are formed, and genes are exchanged between the two chromosomes. Since nonhomologous parts do not pair, there is no exchange of segments between them and no chiasmata are formed, but the presence of large nonhomologous regions does not prevent pairing in homologous parts.

Sex Chromosomes in Plants

Most seed plants are monoecious — that is, both sexes are present on each plant. There is no sex chromosome mechanism, and sex is not a problem of heredity but one of differentiation during development. In some species of plants, however, each individual is either male or female. In some of these

plants a sex mechanism has been discovered like that in animals. In Lychnis dioica, G. H. Shull showed from genetic grounds that the male was heterogametic. Definite X and Y chromosomes have been found in Elodea canadensis, Melandrium album (which is partially synonymous with Lychnis dioica

since it was included in ShulFs L. dioica), hops, poplar, and other plants (Fig. 24) ; inheritance in these plants is of the Drosophila type. One seed plant, Fragaria elatior, is of the Abraxas type. In species of the dock, Rumex, the male is heterogametic for sex, but has two small Y chromosomes and one large X. The two Y chromosomes separate from the X at meiosis, and a male gamete with two Y's produces a male on fertilization whereas one with the one X produces a female.

Fig. 24. Metaphase of first meiotic division in a diploid male plant of Lychnis (Melandrium). Eleven pairs of autosomes are present and one pair which consists of the X and Y chromosomes. Photomicrograph X1400. (Courtesy of Dr. H. E. Warmke in the American Journal of

Botany.)

[Begin Page: Page 74]

Salivary. Gland Chromosomes

After fertilization in Drosophila, the egg is laid in moist food and proceeds to hatch out into a small, crawling larva. Inside the larva, extending back from the mouth a distance of one-fourth to one-third the length of the entire body, are two large

Fig. 25. Chromosomes in a cell of the salivary gland of Drosophila melanogaster. (Courtesy Dr. B. P. Kaufmann.)

salivary glands. These glands have cells so large that they can easily be seen with the low powers of a dissecting microscope. The nuclei of these cells are much larger than those of ordinary cells, being generally about 25 ju, in diameter, and the chromosomes in the nuclei are so large that they are 50 to 200 times as large as the chromosomes in the reproductive cells or in the ordinary body cells of this organism (Fig. 25). Such large chromosomes are characteristic of the salivary glands, the rectal epithelium, and the Malpighian tubules of the entire group, the

Diptera, to which Drosophila belongs. They were first observed in 1881 by Balbiani in the related organism, Chironomus. Their

[Begin Page: Page 75]

possibilities as a tool for studying genetics were not realized for a long time, and during the end of the last century and the early part of the present century they were merely regarded as a puzzling curiosity of no known significance or importance. A little over fifty years after discovery these large chromosomes were correctly interpreted by Heitz and Bauer in Bibio hortulanus and by Painter in Drosophila, and since then they have been studied very intensively.

In addition to being much larger than ordinary chromosomes, the chromosomes in the salivary gland nuclei are atypical in several other respects. Although the nuclei in which these chromosomes are found are not dividing and will not divide again, the chromosomes are not in a typical resting stage but appear to be in a permanent prophase stage with the two homologues of each pair of chromosomes closely paired throughout their length. The pairing of homologous somatic chromosomes is certainly not general but is common in the Diptera, where it occurs in other somatic cells as well as in salivary gland cells. The salivary gland chromosomes are therefore mitotic prophase

chromosomes which have uncoiled and lost the "relic coils" of the previous division and which show a marked somatic pairing.

Striations and Bands

Ordinary somatic chromosomes consist of one or two thin chromonemata or gene strings, but the number of chromonemata in these giant chromosomes is considerably greater. In typical somatic chromosomes, the division of the chromonemata is shortly followed by the division of the chromosome itself, so that the number of chromonemata in a chromosome is never large. In the salivary gland chromosomes of the Diptera, the chromonemata divide a number of times, but these divisions are not accompanied by division of the chromosome as a whole. The result is that a number of these fine threads will be embedded in a common matrix. The number varies with different members of the group. In Drosophila it appears to be about 64, but in other members of the Diptera the number is larger. In Chironomus it is about 400. These chromonemata are completely uncoiled and lie parallel and close to one another throughout the length of the chromosome. They are not uniformly thin,

[Begin Page: Page 76]

but contain a large number of deeply staining chromomeres,

some large, some small, arranged so that the threads appear like strings of loosely strung beads of different sizes. Since all the chromonemata within a common matrix arose from one original chromonema by successive divisions, they should be alike. Apparently they are, for the chromomeres on one chromonema are identical in size and position with the chromomeres on the sister chromonemata. All the chromonemata have a chromomere at exactly the same place on the thread. Since the chromonemata are very close to one another, the chromosomes are in contact laterally. An aggregation of identical chromomeres on the numerous chromonemata appears as a cross-band or disc at right angles to the long axis of the chromosome. One of the outstanding features of these giant chromosomes is the presence of these numerous cross-bands which differ in thickness and in staining capacity and are at various distances apart. Their properties depend upon the size and nature of the chromomeres of which they are composed.

One other feature of the bands is of great genetic significance. In size, position, and sequence the bands in one chromosome are identical with those in the homologous chromosome. Therefore, when the two homologues are paired, as they always are in the salivary glands, the bands of one must lie exactly alongside the corresponding bands of the other. This is strictly true. Pairing of two homologous salivary gland chromosomes is very precise, just as it is in zygotene of meiotic chromosomes. This is so universal a rule, that if a piece of one of the paired homologues involving several bands is deleted by X-rays, the bands of the

other homologue corresponding to those deleted have no bands with which to pair and form a loop to one side. The missing bands in no way alter the pairing of the bands which are present in both homologues. One of the valuable features of these giant chromosomes is the evidence they give of the nature of chromosome pairing.

Bands and Genes

The bands appear to contain a large amount of nucleic acid, whereas the nonstaining or lightly staining regions between the

[Begin Page: Page 77]

bands appear not to contain so much nucleic acid. The many chromonemata that make up the salivary gland chromosome are composed of bundles of fibers of complex organic chemical substances known as polypeptids. Part of these bundles of polypeptid fibers attract nucleic acid, and the remaining parts do not. The parts that contain the nucleic acid stain deeply and form the chromomeres. The fusion of such adjacent deeply staining regions produces a band. If all the chromomeres do not fuse laterally, the band appears as broken. The exact relation between the bands and genes cannot easily be determined. By means of X-rays, certain flies can be produced which have a

marked notch in the wing. When the salivary glands of larvae from such flies are examined, one of the bands is usually seen to be missing from one of the chromosomes, although not from its homologue. The many examples of such notch-winged flies that have been found point to the conclusion that every locus corresponds to at least one band. The bands are therefore in some way correlated to genes although it cannot be said definitely that a band is a gene.

The Chromocenter

Salivary gland chromosomes exhibit one peculiarity not found in other chromosomes. It has been shown that the Y chromosome is made up largely of heterochromatin and that the X chromosomes and autosomes have heterochromatic material around their centromeres although they consist mostly of euchromatin. This condition has little effect upon their behavior in meiotic or in ordinary somatic cells, but in the cells of the salivary gland of Drosophila it has a striking effect. In these cells, all the heterochromatic material of all the chromosomes is fused into a mass from which the euchromatic material extends like tentacles. The entire chromatic material appears like an octopus, with a heterochromatic body and five long and one short euchromatic arms. The long arms are the right arm and left arm of both the V-shaped second and third chromosomes and the rod-shaped X chromosome, and the very small fourth chromosome makes up the sixth projection. In the female, all six arms are of uniform thickness for each consists of two paired homologues. In the male, the X chromosome, since it is an unpaired structure,

[Begin Page: Page 78]

is noticeably thinner than the corresponding chromosome of the female and is thinner than the autosomes of both sexes. The

Y chromosome of the male forms part of the chromocenter. The chromocenter is not a characteristic of dipteran salivary

gland nuclei, although it is a prominent and characteristic feature of Drosophila. In forms like Chironomus and Sciara there is no such union of heterochromatic material, and the chromosomes are independent units in the same number as in typical somatic cells.

"Lampbrush" Chromosomes

Another unusual type of chromosome has recently received considerable attention. In some animals, including amphibians and birds, during the maturation of the egg, the chromosomes of the first meiotic prophase may increase greatly in length. This increase occurs in those forms whose developing eggs remain in the prophase for a considerable time. The homologous chromatids pair normally, but their chromonemata then proceed to become extended by uncoiling As this continues, outgrowths

which are very fine threads appear from the sides of the chromonemata. They grow out approximately perpendicular to the chromonemata and then bend around into loops. These loops always appear in a certain order on the main thread, being of a characteristic number and a characteristic distance apart from one another. It has been suggested that they give forth substances into the cytoplasm which exert an action in directing the development of the egg, but they need more study for a completely satisfactory explanation of their behavior.

QUESTIONS AND PROBLEMS

1. Show by diagrams how the X chromosome and the Y chromosome would pair at meiosis if the X chromosome were rod-shaped and the

Y chromosome were J-shaped if: (a) the long arm of the Y chromosome were completely homologous with the X chromosome; (6) the proximal part of the long arm of the Y chromosome were homologous with an interstitial segment of the X chromosome; (c) there were no homologous parts in the X and Y chromosomes.

2. Discuss the possibihties (a) that maleness is determined by the presence of a Y chromosome and (6) that sex is determined in a diploid animal by either one or two X chromosomes.

[Begin Page: Page 79]

3. Does the presence of two X's or of an X and a Y have any effect on the somatic divisions of an organism ? If so, what is the effect ?

4. By a series of diagrams show how the formation of many new chromonemata within a common matrix could produce banded structures such as are found in sahvary gland chromosomes.

5. What is the significance, if any, of the chromocenter? Is it an essential feature of salivarv gland nuclei?

[Begin Page: Page 80]

Chapter 6

THE GENETIC DISTRIBUTION OF A PAIR OF ALLELES LOCATED IN AUTOSOMES

It has been shown that plants and animals may be homozygous for a dominant or for a recessive gene or that they may be heterozygous. For example, an evening primrose plant may be homozygous for the recessive gene, bullata, and have short, crinkled leaves, or it may contain the dominant allele, in which case it will have noncrinkled, or normal, leaves. In the four-o'clock, plants which are homozygous for the gene for red will have the

character, red flowers, and those homozygous for white, the allele of red, will have the character, white flowers. As these genes do not exhibit dominance, the character shown by the heterozygote is pink flowers. Obviously, it is the gene and not the character that is transmitted from generation to generation since germ cells do not have such structures as leaves and flowers. The method by which the genes are distributed is one of the most important and best-understood problems of the science of genetics.

Since the genes are located in the chromosomes, the problem of the distribution of genes is inseparable from the problem of the distribution of the chromosomes. The behavior of the chromosomes in the formation of spores, gametophytes, gametes, and zygotes has already been pointed out. The next step is to study the behavior of a pair of chromosomes which contain a certain known pair of alleles. In the second chromosome of Drosophila melanogaster the locus of c, the gene for curved wings, is found about three-fifths of the distance (genetically) from the end of one arm. The other five hundred odd genes in this species can be ignored for the present and observation can be limited to gene c and its allele, C. In cooperation with a large number of non-allelic wild-type genes, C produces normal wings. When only one pair of genes is under consideration, the situation is simple. Organisms which are heterozygous for only one pair of alleles or

[Begin Page: Page 81]

organisms in which only one pair of alleles is being studied are called monohybrids. Some interesting characters in maize determined by single genes are shown in Fig. 26, and a striking structural character in Shepherd's-purse in Fig. 27.

Fig. 26. Some interesting inherited types in Zea mays. Left to right, pop, flint, sweet, pod, and dent. The sweet and floury types appear to differ from flint by single recessive genes, and the pod type by a dominant, but popcorn and dent probably differ by a number of interacting genes. (Photograph by Dr. W. Brooks Hamilton.)

If a fly is homozygous for curved wings, it will have two c genes, one in each homologue. Since the two homologues separate at meiosis the two c genes must separate, or segregate, also. Since each gamete has only one member of chromosome II, it can have only one c gene; and since the fly is homozygous, all its gametes must be alike. Similarly, in flies homozvgous for the dominant allele, all the gametes must have gene C and must have only one

such gene, for meiosis operates in the same manner in a dominant as in a recessive. In the heterozygote, one chromosome bears C and the homologous chromosome bears c. Since these chromosomes separate during meiosis and enter different gametes, every gamete must have C or c, but never both. Theoretically, exactly half the gametes of the heterozygote would have C and exactly half would have c. The two genes at a given locus segregate jrom

Fig. 27. A single gene difference in Shepherd's-piirse : left, a rosette of Capsella (Bursa) grandiflora and, right, a rosette if C. Viguieri of the same age. The latter type possesses a dominant gene which produces a heavy fasciation of the stem.

one another at meiosis; as a result, only one member of the pair of alleles is present in each gamete. This separation of the two genes at any locus is the principle involved in MendeVs first law, often called the law of segregation. The behavior of the genes in the formation of gametes and plant spores is merely a function of the behavior of the chromosomes.

The genetic constitution of an organism is the result of the particular gametes which happen to unite when that individual is formed. For example, if an egg bearing C is fertilized by a sperm

which has the C gene, a homozygous normal-winged fly is produced. If both the egg and sperm happen to contain the gene c, the resulting individual will have curved wings. If the egg is C and the sperm c, or if the egg is c and the sperm C, the new fly will be genotypically heterozygous and phenotypically wild type.

[Begin Page: Page 83]

It can be seen, therefore, that all the offspring of a homozygous dominant will be phenotypically dominant irrespective of the genetic constitution of the other parent, since all the gametes of the homozygous dominant would contain the dominant gene.

If a homozygous wild type is crossed with a curved, all the offspring will be heterozygous and will look like the dominant parent. The generation of the parents is called the parental, or Pi, generation; the generation of the offspring is known as the first filial, or Fi, generation (pronounced "eff one"). Since all the Fi flies are alike genotypically, all will produce the same kinds of gametes. Since all are heterozygous, one half the gametes of each fly will be C and the other half will be c. If two Fi flies are mated together, the grandchildren of the original two parents will be produced. This generation is known as the second filial, or F2, generation. One half the eggs of the Fi fly which is used as a female will contain C and one half will contain c. Simi-

larly, the male Fi fly wdll produce C and c sperm in equal numbers.

Whether a C-bearing sperm unites with a C or with a c egg is a matter of pure chance as the probabilities are theoretically even. The same is true for the sperm which bear c. The chance that a C egg will be fertilized by a C or by a c sperm is exactly even, and the same is true for a c egg. Therefore, four combinations are possible in the F2 and will exist in equal numbers: CC, Cc, cC, and cc. Since Cc and cC flies are alike genotypically, the nature of the F2 can be written as a ratio of ICC : 2Cc : Ice. In terms of fractions, the F2 population will be y4:CC : Y2CC : y4cc. These ratios, however, are genotypic. As both homozygous C and heterozygous flies are phenotypically alike, the phenotypic F2 ratio is 3 normal-winged : 1 curved or % normal and % curved. This is shown diagrammatically in Fig. 28, in which the method of arriving at the F2 is determined by the conventional "checkerboard." It must be understood that if only four flies are produced, three of them will not necessarily be normal and one curved. This ratio is theoretical, based on chance, and means that out of a large number of cases, approximately three-quarters will be normal and approximately one-quarter will be curved. The larger the number of F2 flies, the more nearly the numbers obtained may be expected to approximate the theoretical ratio.

[Begin Page: Page 84]

Monohybrids

P, flies

Pi gametes

F, flies

Fi gametes

F, flies

Homozygous Wild Type Curved

CC cc

/\ /\

© ©. © ©

Heterozygous

Cc

/\

© ©

Fi sperm

C c

F.

Eggs

C

c

F2 genotypic ratio

F2 phenotypic ratio

ICC 2Cc : Ice

3 Wild Type: 1 Curved

Backcross of Fi to

Dominant Parent

C C ^ Cc

Backcross of Fj to

Recessive Parent

c c ^ Cc

All Wild Type

1 Wild Type: 1 Curved

Fig. 28. The checkerboard method of determining the F2 and backcross generation.?. A cross between a wild-type female {CO and curved winged male {cc) produces a heterozygous wild type {Cc). The eggs of the Fi are C or c as are the sperm. Since either kind of sperm can unite with either kind of egg at random, four possible kinds of F2 individuals will be produced in equal numbers; but since two kinds are alike, the Fo genotypic ratio will be ICC : 2Cc : Ice. Since dominance is complete, the F2 phenotypic ratio will be 3 wild type : 1 curved. The backcrosses of the Fi to the dominant and to the recessive parents are indicated in the lower half of the figure.

Backcross

If an Fi fly is mated with one of its own parents, the cross is called a backcross. If the Fi is backcrossed to the dominant parent, all the ofTspring will be alike phenotypically although genotypically half will be CC and half Cc. If the backcross is made with the recessive parent, one half the offspring will be genotypically Cc and the other half will be homozygous for the

c gene. The phenotypic ratio when the Fi is backcrossed to the recessive parent is 1 wild type : 1 curved. The 1 : 1 ratio will be obtained whenever any heterozygote is crossed with a homozygous recessive. ''Backcross'' is literally appropriate only when an Fi animal or plant is crossed with one of its own parents. At other times ^'testcross'' is more appropriate, although the terms are frequently used indiscriminately.

The cross of a heterozygote with a recessive will give a 1 : 1 ratio whereas a cross between a homozygous dominant and a recessive will produce only dominants. Use can be made of these facts to test whether a dominant plant of unknown ancestry is homozygous or heterozygous. In certain varieties of lupines, red flowers are dominant over white. If a commercial seed house wishes to market seeds of a red variety of lupine, claiming that only red-flowered plants will be produced, and if they have a number of red-flowered plants from which to obtain their seeds, they must know the genotypes of the plants before they can market seeds from them with a guarantee that all will yield plants with red flowers. If the red-flowered plants used for seed are homozygous, all the seed from them will produce red-flowered plants; but if some are heterozygous, one-fourth of the seed from those plants will produce plants with white flowers.

If the company guarantees the seeds to produce only red-flowered plants, it must know which of the plants are heterozygous and which will breed true for red flowers. One of the most widely used methods of testing them is to cross them with recessive, white-flowered plants. Those plants which produce only red-flowered offspring, when mated with recessives, are the homozygotes and are used to produce the red-flowered seed for the market. Those, on the other hand, that give approximately equal numbers of red- and white-flowered plants when crossed with the recessive are heterozygotes and worthless for this particular purpose. A cross of a dominant of unknown genotype with the recessive is a widely used method of determining the genotype of phenotypically dominant plants and animals.

Practical Considerations in Using the Testcross. It is obvious that the method of the testcross is not the only way of determining whether a plant is homozygous or heterozygous. Self-fertiliz-

[Begin Page: Page 86]

ing a plant and raising the offspring would accomplish the same result. To be certain of including a sufficient number of recessives to establish the nature of the unknown within the realm of probability, however, the seedsman would have to grow a much larger number of plants from a selfing than from a testcross.

The use of seed from a self-pollination would require more land than seed from a testcross, and that land might be used more profitably for another purpose; it also would require more labor to pot and set out the additional plants required by this method. Usually, therefore, the testcross method is more practical from an economic viewpoint. However, the best method to use is also determined in part by the nature of the plant under consideration. Each flower of wheat produces one seed. This plant is self-fertihzed with no difficulty, but the labor involved in making over a hundred hand pollinations is a great item of expense. For wheat, the expense of making the crosses might outweigh that of the additional land and labor necessary for a test by self-fertilization and might make testcrossing impractical. The situation would be different for a plant like tobacco where one hand pollination would produce several hundred seeds; for it the testcross method would be more desirable.

Species of animals in which the individuals are of one sex only are tested by the testcross method as it is obviously impossible to self-fertilize them. It would be possible to test an unknown dominant animal by crossing with a known heterozygote, but such a method would be no simpler than to cross with a recessive and would require a greater number of offspring so as to be sure to include a reasonable number of recessives.

Incomplete Dominance

When a homozygous dominant is crossed with a homozygous recessive, the Fi is phenotypically like the dominant parent and

the F2 splits into three dominants to one recessive if dominance is complete. When dominance is incomplete, however, the Fi does not resemble either parent and the phenotypic ratio in the F2 is identical with the genotypic ratio. In the four-o'clock, a red-flowered plant, WW, crossed with a white-flowered plant, WW, would give a pink-flowered Fi, Ww; this, when selfed, would show a segregation in the F2 into 1 red (WW) : 2 pink (Ww) : 1 white (ww).

[Begin Page: Page 87]

Another striking case of incomplete dominance is the often-cited blue Andalusian fowl. This variety is a heterozygote and can be produced only by crossing a black with a white. There is no dominance, and the Fi from such a cross is neither black nor white but a peculiar intermediate shade called "blue" (Fig.

Fig. 29. A blue Andalusian female. This t3^pe is the heterozygote from a cross between certain black and white types. Therefore it never breeds true when crossed with a blue Andalusian male, but produces a ratio of 1 black : 2 blue : 1 white. In the female the neck feathers are dark blue. In the male the back and saddle feathers, in addition to the neck feathers, are almost a solid blue. (Courtesy of Dr. M. A. Jull.)

29). Since the blue fowls are always heterozygotes, they cannot possibly be made to breed true. When two blue fowls are mated, the offspring are in the ratio of 1 black : 2 blue : 1 white. When a blue is mated with a white the offspring are 1 blue : 1 white, and when a blue and a black are crossed the resulting ratio is 1 black : 1 blue.

An interesting dominance relationship is found in certain crosses between horned and hornless sheep. In Dorset Horn sheep, the males have very large horns and the horns of the

[Begin Page: Page 88]

females are smaller. If Dorset sheep are crossed with a hornless breed such as the Suffolk, the Fi females are hornless, but the males have horns, although these horns are considerably smaller than those in the pure Dorset Horn breed. Dominance of hornlessness is complete in the females and incomplete in the males.

Reversal of Dominance

If dominance is complete, the dominant character occasionally may develop more slowly in the heterozygote, in which there is

only one dominant gene, than in the homozygote, in which two dominant genes are present. Shepherd's-purse, Capsella bursa-pastoris, is a good example of this. A gene, A, produces sharp leaf lobes, whereas its allele, a, produces rounded lobes. If a plant homozygous for A is crossed with a recessive, the Fi plants resemble the recessives when young, but their leaf lobes gradually elongate and become pointed as they mature. In the mature condition the heterozygous Fi plants are unmistakably like the homozygous dominants. In the F2 when young plants are examined, the ratio appears to be 1 pointed lobe : 3 round lobes, but in the adult condition the ratio changes to 3 pointed : 1 round, as the dominant gene in the heterozygote comes into expression. The gene which produces the elongation of the lobes acts more slowly when present in only half quantity, but the mature heterozygotes are indistinguishable from the homozygous dominants.

Reciprocal Crosses

It is conventional in writing crosses (except in human genetics) to write the female first and the male second. The cross, wild type X curved, means that a normal-winged female is mated with a curved-winged male, and curved X wild type means that a female with curved wings is mated with a normal-winged or wild-type male. The cross curved X wild type is known technically as the reciprocal of the cross wild type X curved. For genes on autosomes the results are generally the same no matter in which direction the cross is made. The Fo of the cross

curved X homozygous wild type segregates into 3 wild type and 1 curved just as did its reciprocal. In testcrosses, also, the results are the same no matter in which direction the cross is